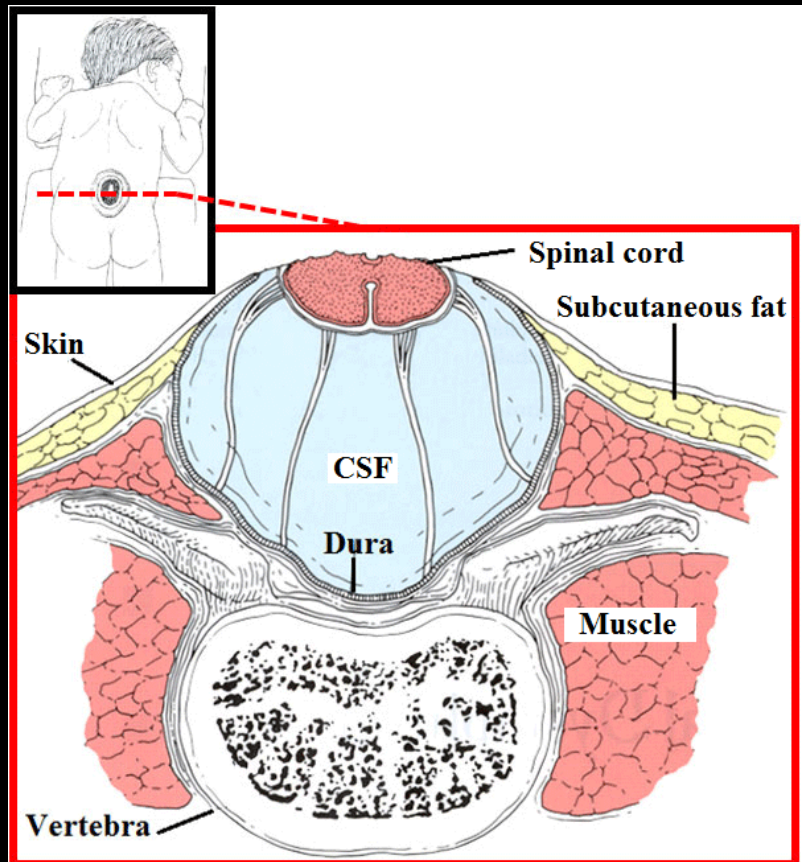
I disrafismi spinali

Francesco Sala

SSO di Neurochirurgia Pediatrica, Istituto di Neurochirurgia
Azienda Ospedaliero-Universitaria Integrata
Verona

Spina bifida aperta

- Mielomeningocele

Spina bifida aperta

- Mielomeningocele: screening ecografico prenatale
- l'accuratezza di tale indagine raggiunge il 75%
- in un altro 18% dei casi la diagnosi di "spina bifida" è corretta in termini di riconoscimento del difetto, ma imprecisa nell'esatta definizione del tipo di lesione
- nel restante 7% dei casi la malformazione non è stata riconosciuta
- Questi dati impongono cautela nell'affrontare con la famiglia del feto malformato il problema della prognosi e dell'eventuale interruzione della gravidanza

Spina bifida aperta

- Mielomeningocele
- Chiari II
- Idrocefalo

Spina bifida occulta

- Disrafismi spinali con cute integra
- Non associati a Chiari II nè a idrocefalo
- Spesso associati ad anomalie cutanee della linea mediana
- Si presentano clinicamente con una sindrome da midollo ancorato

Arch Dermatol. 2004;140:1109-1115

STUDY

Skin Markers of Occult Spinal Dysraphism in Children

A Review of 54 Cases

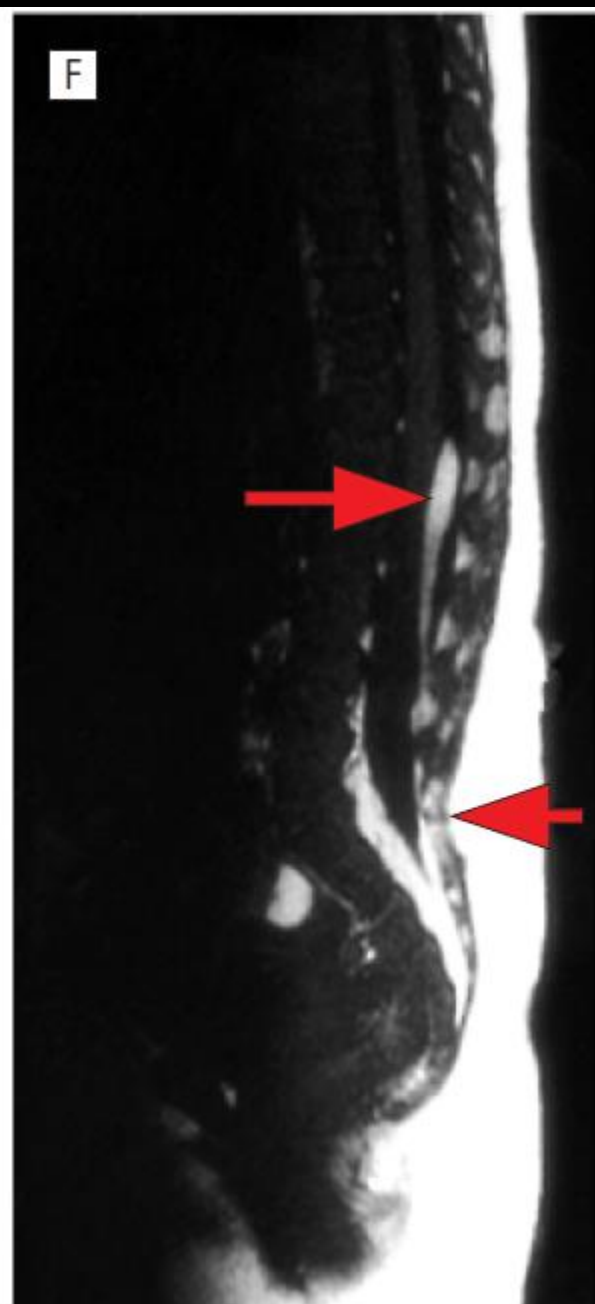
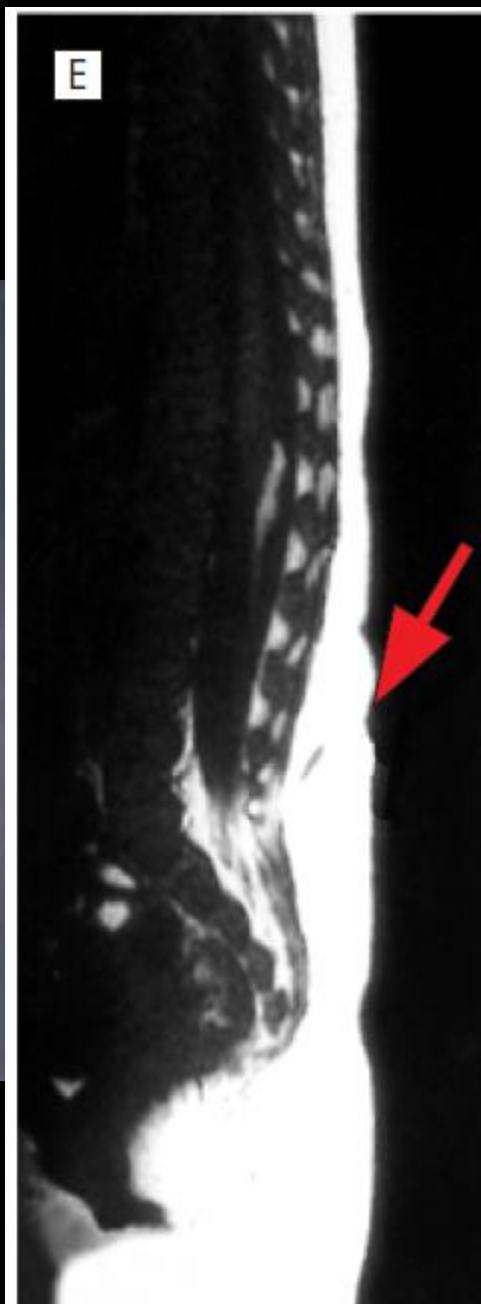
David Guggisberg, MD; Smaïl Hadj-Rabia, MD; Caroline Viney, MD; Christine Bodemer, MD, PhD; Francis Brunelle, MD; Michel Zerah, MD, PhD; Alain Pierre-Kahn, MD; Yves de Prost, MD; Dominique Hamel-Teillac, MD

Results: Occult spinal dysraphism was detected in 3 of 36 patients with an isolated congenital midline lesion and 11 of 18 patients with a combination of 2 or more different skin lesions.

Conclusions: A combination of 2 or more congenital midline skin lesions is the strongest marker of OSD. Careful dermatologic examination is needed to detect suggestive markers and request a spinal magnetic resonance image, which is the most sensitive radiologic approach to detect an OSD.

A
D

B
E
F

C
G

Spina bifida occulta e anomalie cutanee

- La spina bifida occulta si associa frequentemente ad anomalie cutanee della linea mediana (midline skin sigmata=MSS)

- La maggior parte degli infanti con MSS non hanno una spina bifida occulta

Diagnostica

- Ecografia:
  - Non invasiva, accessibile
  - Operatore dipendente, limitata dall'ossificazione vertebrale
- RMN:
  - Gold standard, accurata
  - Costosa, richiede anestesia generale

Conclusioni

- L’ecografia ha dimostrato eccellente sensibilità (96%), specificità (96%) e valore predittivo positivo (96%)
- Viene consigliata come esame di screening per “isolated low risk MSS” (fossette sacro-coccigee, seno pilonidale, deviazioni della piega glutea)
- Le dimensioni del seno, la sua profondità e la traiettoria non sono considerate rilevanti
- E’ fondamentale l’esperienza dell’ecografista!

ORIGINAL ARTICLES

Low-Risk Lumbar Skin Stigmata in Infants: The Role of Ultrasound Screening

Liat Ben-Sira, MD, Penina Ponger, MD, Elka Miller, MD, Liana Beni-Adani, MD, and Shlomi Constantini, MD, MSc

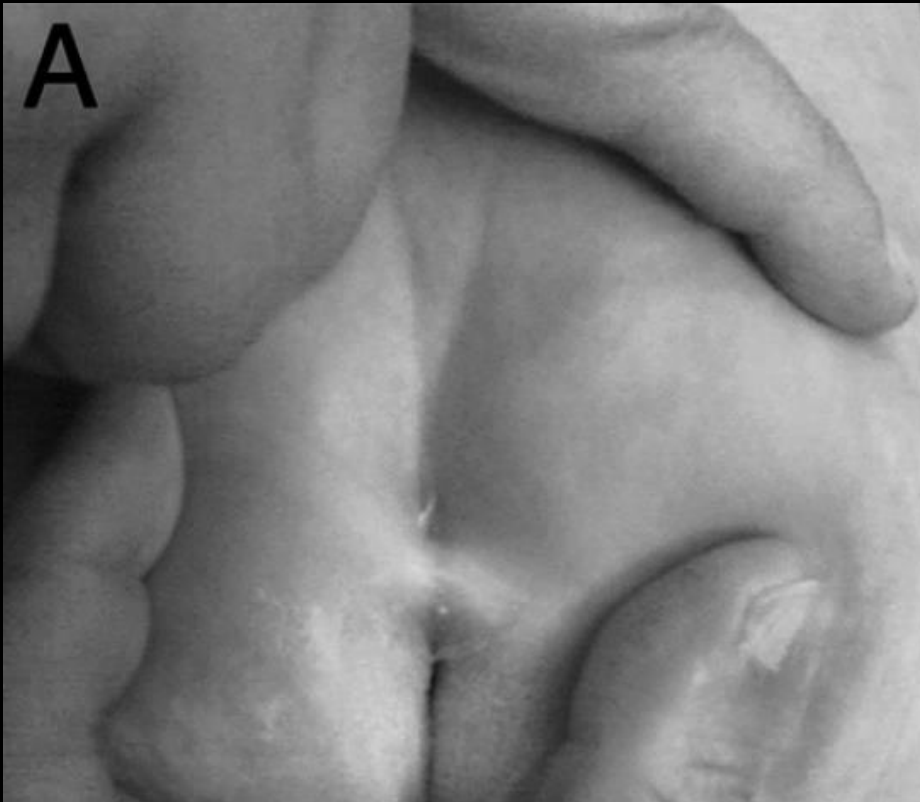
Objective To reassess the utility and validity of ultrasound (US) screening in infants with lumbar midline skin stigmata (MSS) that may be associated with tethering of the spinal cord.

Study design We conducted a prospective observational study of 254 infants under age 6 months with suspicious dorsal MSS between 2005 and 2007. All infants were examined by US and neurosurgical clinical evaluation, and 50 infants also underwent magnetic resonance imaging (MRI). The US and MRI findings were analyzed for correlation. Associations between the imaging findings and the presence of the low-risk skin lesions simple dimple (113 cases) and deviated gluteal fold (DGF; 44 cases) also were evaluated.

Results Analysis of US and MRI results for the cohort of 50 neonates in whom both examinations were performed showed high concordance. The low-risk group of infants with simple dimple and DGF constituted 157 US procedures, 96% of which were of high quality, providing clear visualization of spinal components. None demonstrated any clinically significant pathological findings.

Conclusions Our data reaffirm the reliability of US as a screening tool for tethered cord syndrome. Infants with low-risk lesions, such as simple dimple and DGF, may be absolved from US screening, because these findings alone do not indicate underlying pathological lesions. We propose a simplified diagnostic classification system for MSS. *(J Pediatr 2009;155:864-9)*.

Diagnostica

- Simple dimple:

Diagnostica

- Deviated gluteal fold:

Diagnostica

- Ecografia:
  - Indicativi di “tethered cord”
    - Cono midollare al di sotto del disco L2-L3
    - Cono midollare non inferiore a L2 ma associato a:
      - Siringe
      - Lipoma
      - Seno dermico al di sopra del “cul de sac” subaracnoideo
      - Filum ispessito

Diagnostica

- Pazienti inviati principalmente per “simple dimple” (n=125) e DGF (n=53)

- Ecografia eseguite in media a:
  - 7 settimane per simple dimple
  - 9 settimane per DGF

- RMN in 50 casi, in media a 20 settimane di vita

In our prospective study, all US examinations were performed by a single senior pediatric radiologist (L.B.S.). The quality of and conclusions that can be drawn from US under such conditions may be strongly operator-dependent. Inexperienced examiners may increase the rate of false-positive and false-negative findings. US of the lumbar spine must be performed at a young age and by a professional imaging expert trained in the field. In addition, open communication between the radiologist and clinician is crucial to effective evaluation.

Fossette sacrali
(simple dimple up to 2.5 cm above the anus)

- 48% nessun esame
- 30% Ecografia
- 22% RMN

Deviazioni piega glutea

- 49% ecografia
- 30% RMN
- 21% nessun esame

L'indicazione all'imaging diagnostico aumenta nel caso di deviazioni asimmetriche, in presenza di una massa palpabile o di altre anomalie cutanee

Emangioma

- 74% RMN
- 18% Ecografia
- 8% nessun esame

Maggior tendenza a studiare emangiomi che risultano ispessiti, scuri, di grandi dimensioni, con superfice irregolare e quelli che si modificano nel tempo

Più di una lesione

- >90% RMN

Radiografie standard?

- X-ray imaging has become an outdated imaging modality, often unreliable as the neonatal skeletal system has yet to complete ossification.
- Spina bifida on x-rays has no clear association with tethered cord

Diagnostica per gruppo di rischio

- Basso rischio (fossette sacrali, DGF, Mongolian spots, peluria): nessun esame o Ecografia

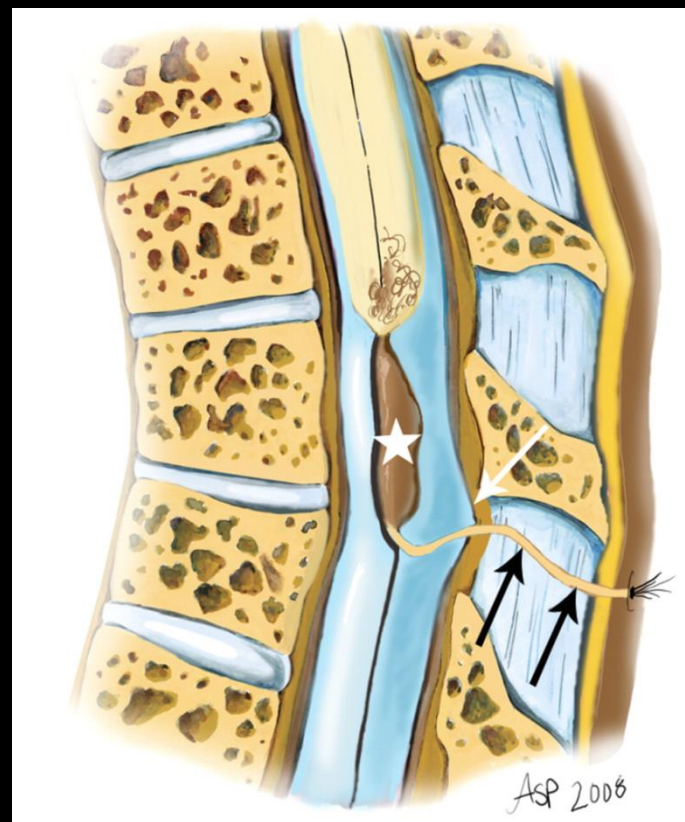
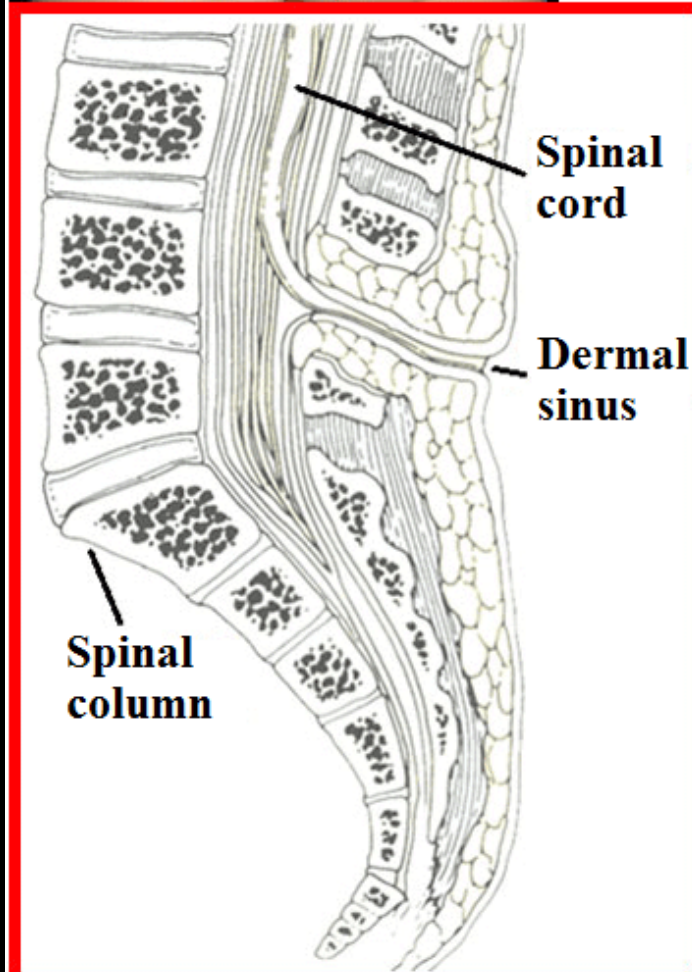
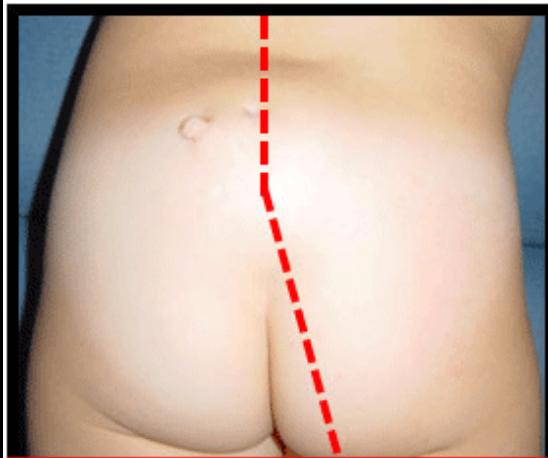
- Rischio intermedio (qualunque discromia cutanea lombare mediana): Ecografia e/o RMN

- Alto rischio (fossette lombari, ipertrichosi, masse, anomalie multiple): RMN

Seno dermico

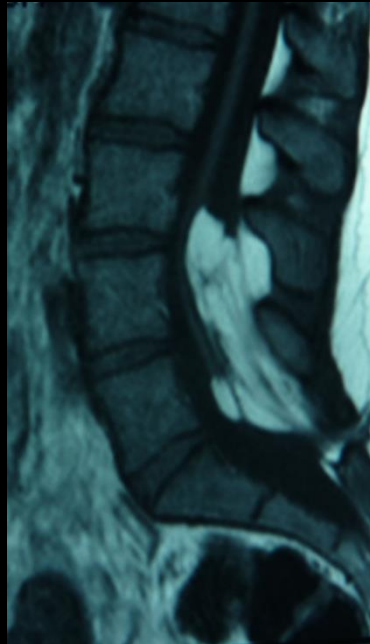
- E’ il risultato di una aderenza anormale dell’ectoderma del tubo neurale al derma. Il tessuto dermico può a questo punto svilupparsi intrappolato all’interno del tratto ectodermico.
- A livello cutaneo, è spesso frequente una fossetta della pelle come pure un emangioma o un’ipetricosi.
- Si estende dalla cute fino ad una profondità variabile. Passa attraverso lo strato sottocutaneo, attraverso la schisi delle lamine verso il sacco durale
- Può anche terminare in un tumore dermoide o epidermoide situato tra le radici spinali della cauda equina.
- Può provocare: ancoraggio del cono midollare, effetto massa quando associato ad un tumore dermoide od epidermoide, e la meningite batterica

ASP 2008

Spinal cord
Dermal sinus
Spinal column

Perché è importante riconoscere la spina bifida occulta?

- Durante la neurulazione secondaria, la colonna si allunga più velocemente del midollo spinale, con il risultato di una progressiva ascesa del cono midollare
- Il cono raggiunge la sede definitiva durante i primi mesi di vita

Tethered Cord

- “Any pathologic fixation of the spinal cord in an abnormal caudal location”

Conus medullaris

Tethered cord syndrome

- Hoffman, Hendrick, Humphreys. The tethered spinal cord: Its protean manifestations, diagnosis, and surgical correction. Child's Brain 1976; 2:145-155.
  - 31 children with spina bifida occulta
  - Low back pain
  - Progressive neurological deficits involving lower limbs
  - Urologic disturbances (incontinence)
  - Orthopedic abnormalities

Pathophysiology of tethered cord syndrome

- Yamada, JNS 54:494-503, 1981
  - Marked metabolic and electrophysiological susceptibility to hypoxic stress of the lumbosacral cord under constant or intermittent traction
  - Untethering improves mitochondrial oxidative metabolism

Adults with TCS

- Patients with previous repair of myelomeningocele
- Patients with progressive symptoms since childhood
- Patients with a stable neurological deficit, foot deformity or scoliosis recognized in childhood but with new or progressive symptomatology in adulthood
- Patients with “occult” spinal dysraphism (but some have cutaneous markers) that remained asymptomatic over several years

Neurosurg Focus 29 (1):E1, 2010

Tethered cord syndrome: a review of the literature from embryology to adult presentation

Dean A. Hertzler II, M.D., John J. DePowell, M.D., Charles B. Stevenson, M.D., and Francesco T. Mangano, D.O.

TABLE 4: Summary of common symptoms in adults presenting with TCS

Authors & Year	No. of Patients	Mean Age at Presentation	% w/ Pain	% w/ Motor Weakness	% w/ Bladder Dysfunction
Rajpal et al., 2007	61	36	56	79	34
Lee et al., 2006	59	43	73	78	71
Huttmann et al., 2001	56	34	77	57	70
van Leeuwen et al., 2001	57	41	74	44	67
Pang & Wilberger, 1982	23	39	78	65	56

Authors & Year	No. of Patients	Back Pain
Pang & Wilberger, 1982	23	100% better
Iskandar, et al., 1998	34	81% better, 15% same, 4% worse‡
Gupta, et al., 1999	18	89% pain "disappeared"
Akay, et al., 2000	10	100%
Yamada & Losner, 2000	10 w/ spinal dysraphism, 60 w/ no history of spinal dysraphism	100% better (patients w/ spinal dysraphism), "most" better (other patients)
Huttmann, et al., 2001	56	91% better, 4% same, 4% worse (at 6 mos)‡
Van Leeuwen, et al., 2001	57	56% better, 39% same, 6% worse‡
		48% better, 45% same, 8% worse‡
Phi, et al., 2004	16 (2 patients had asymptomatic TCS)	14% better, 14% worse, 72% same‡
Quinones-Hinojosa, et al., 2004	13	62.5% better, 25% same, 12.5% worse‡

275 PATIENTS

Pain reduction 86%

Sensori-motor deficits 35-71%

Bladder-bowel dysfunction 16-60%

What determines when symptoms begin is likely how much tension is on the filum terminale

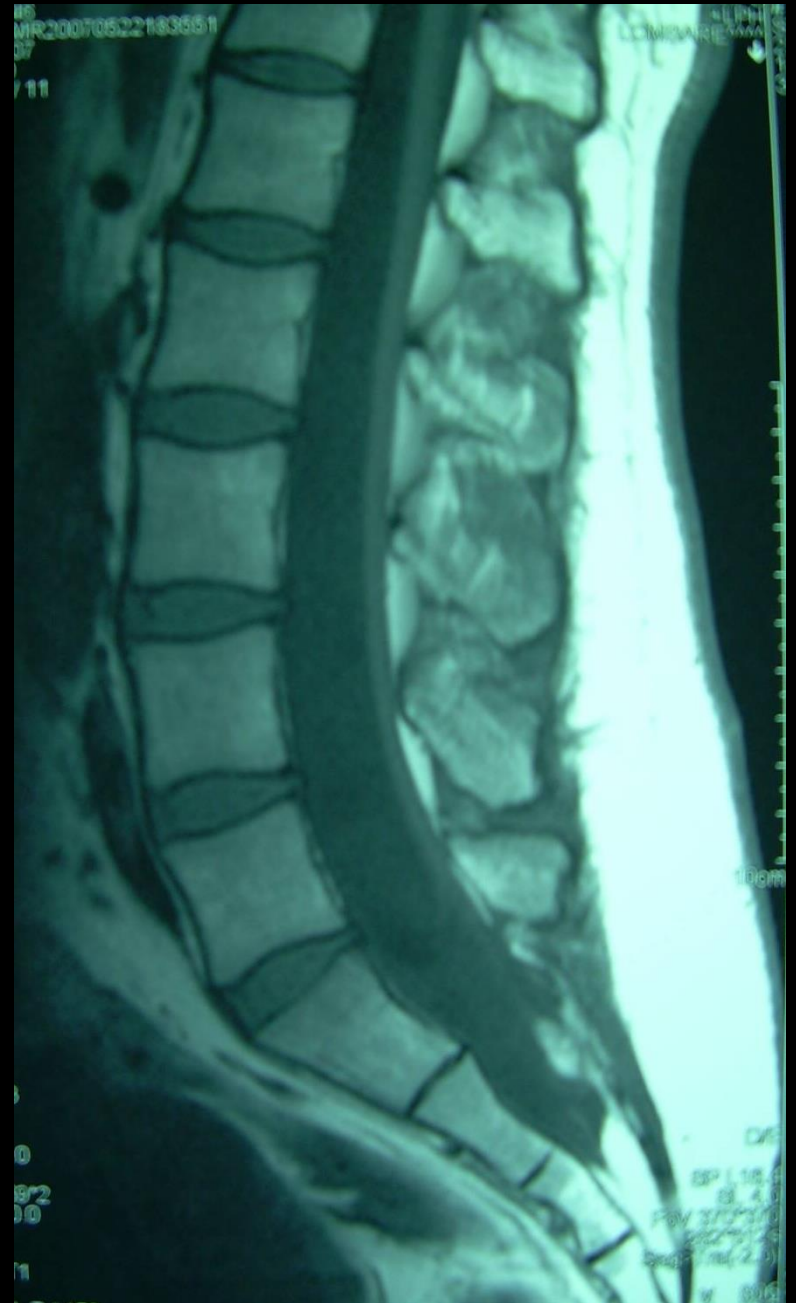
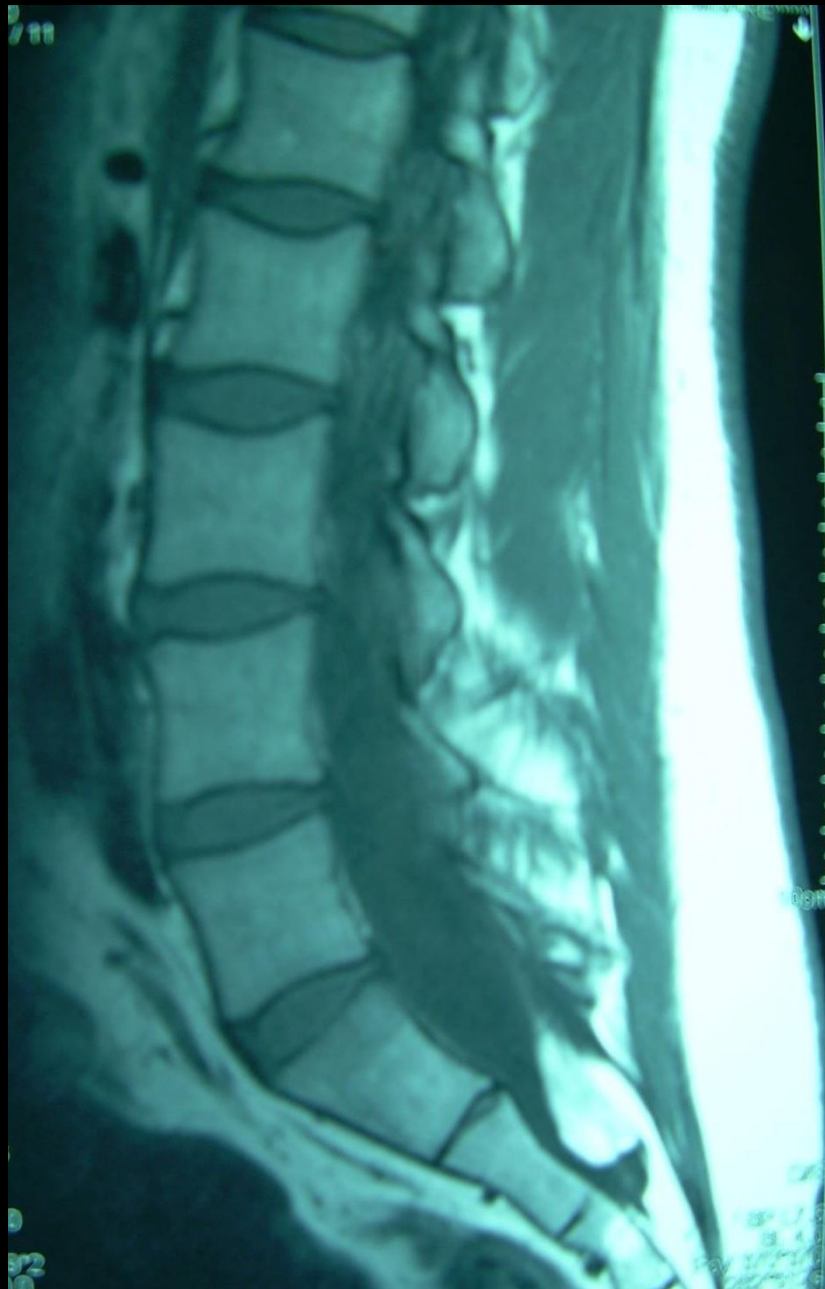
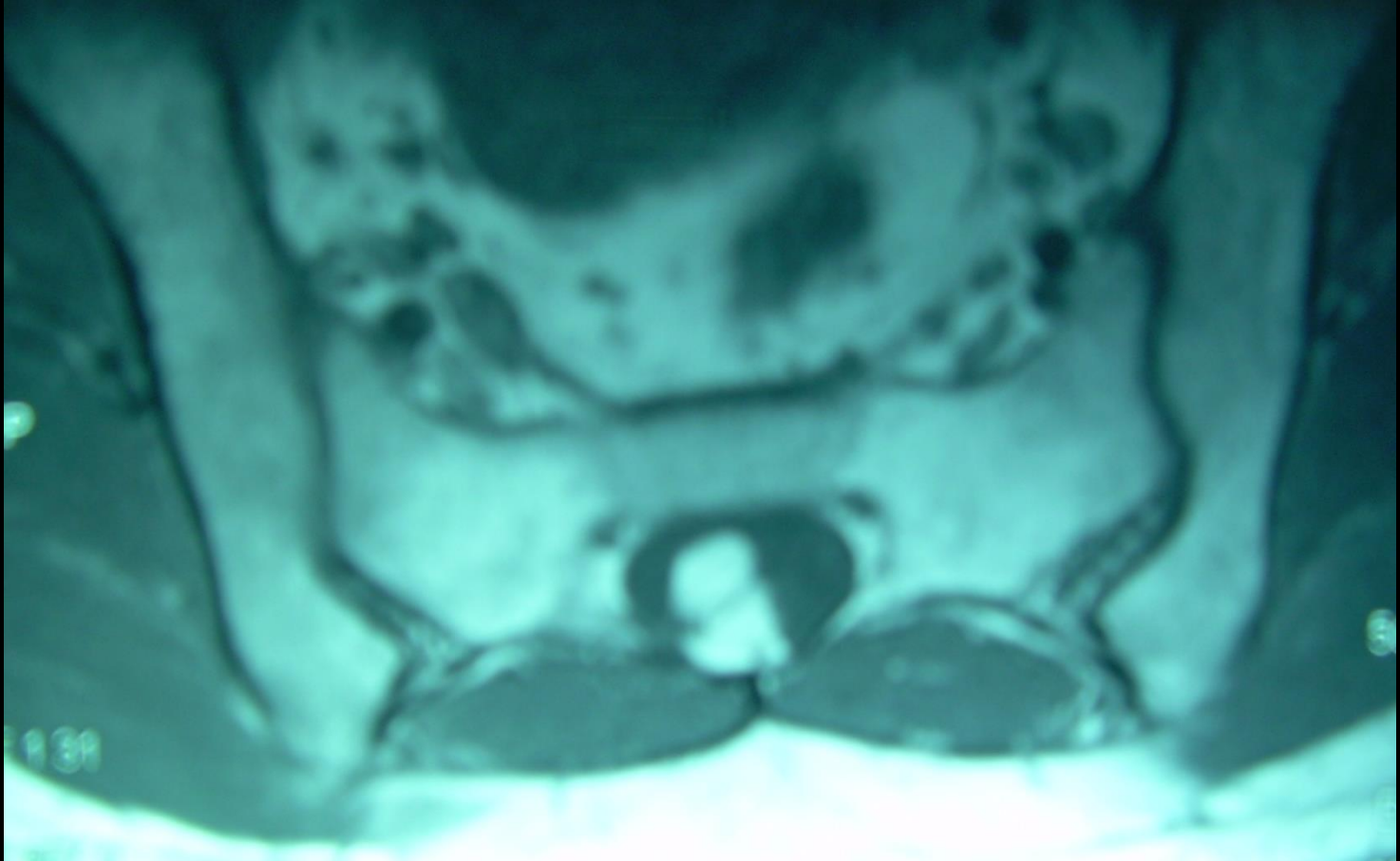
When to operate ?

Natural history of a tethered cord is progressive loss of neurologicl function, over years

(older patients rarely present neurologically intact)

B.A., 36-year old male

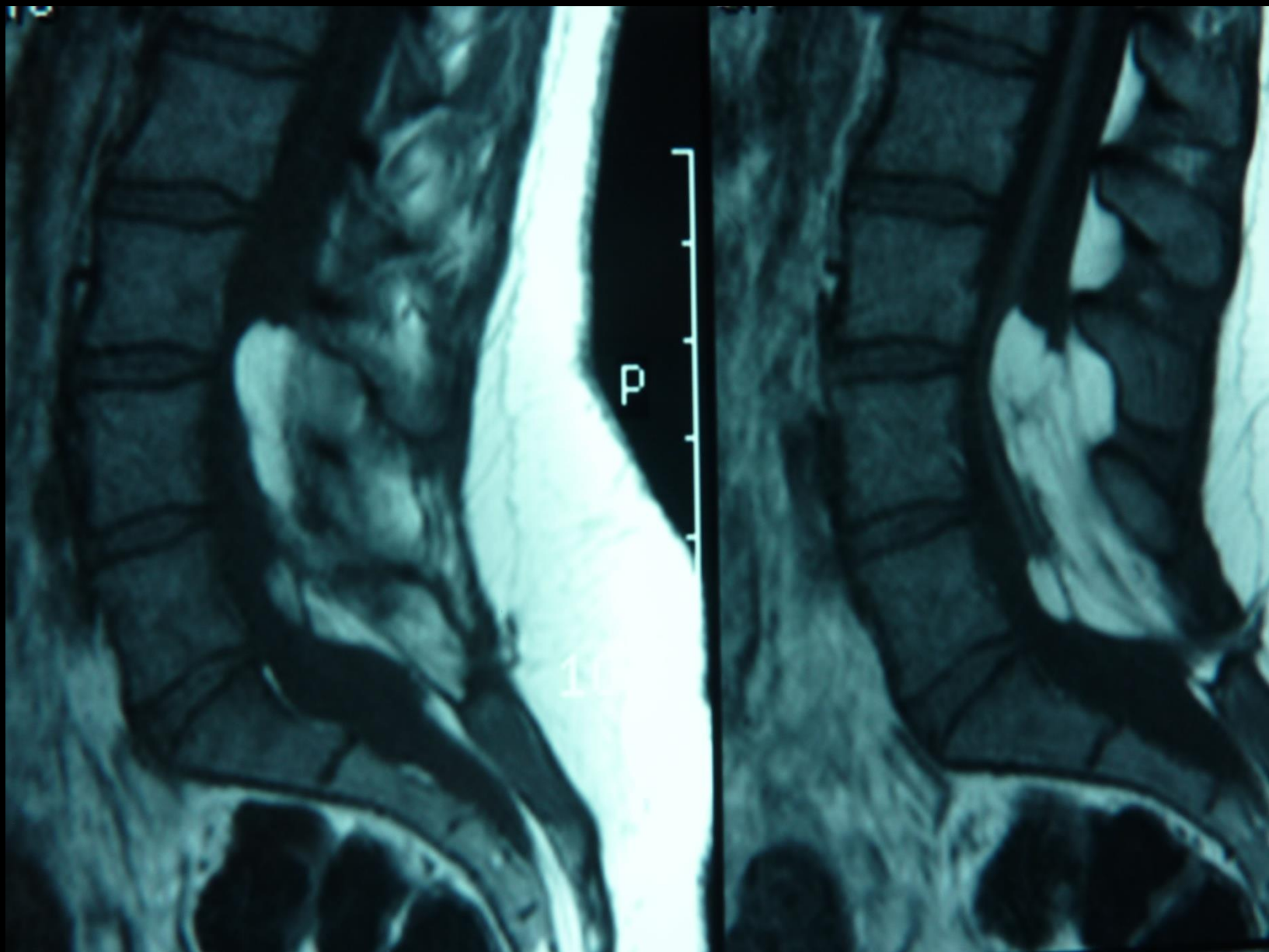
- Pes cavus bilaterally
- At age 22 some mild urinary disturbances that progress to urinary retention
- First MRI 14 yrs. later
- Repeated UTI over the past few months
- Autocatheterization

B.A.

B.A.

Q.A., 25-year old female

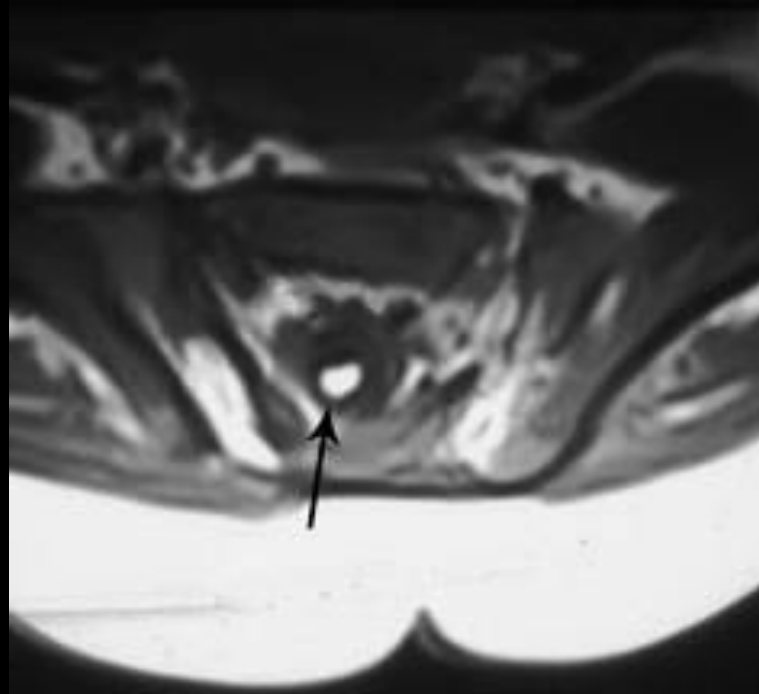
- Operated on twice during childhood for syndactyly on the right foot
- Hairy patch
- Low back pain for several year, worsen since January 2011
- Right leg paresthesia since March 2010
- No bladder bowel dysfunction
- In February 2011 lumbosacral spine Rx, then MRI

Q.A.

Q.A.

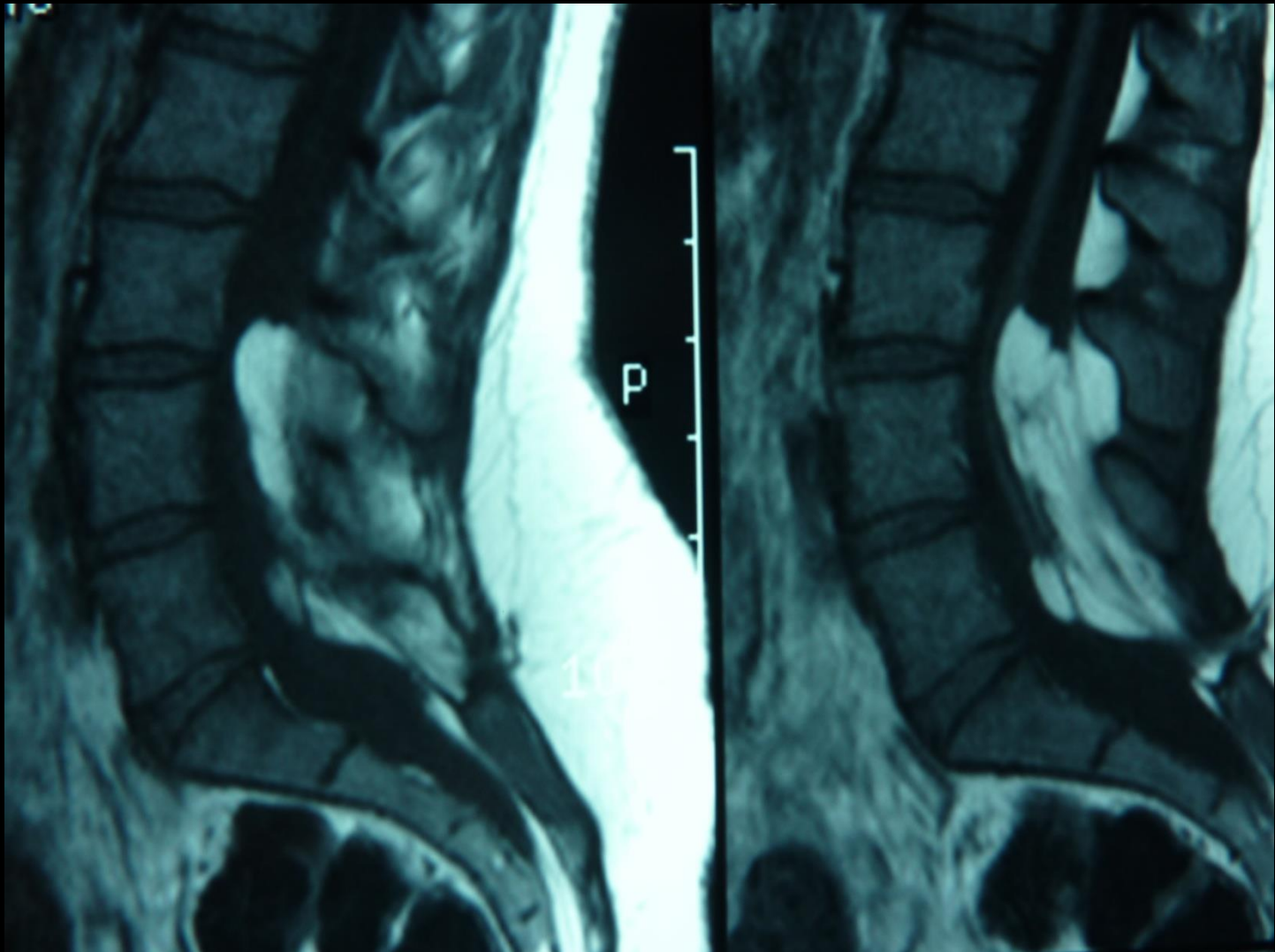
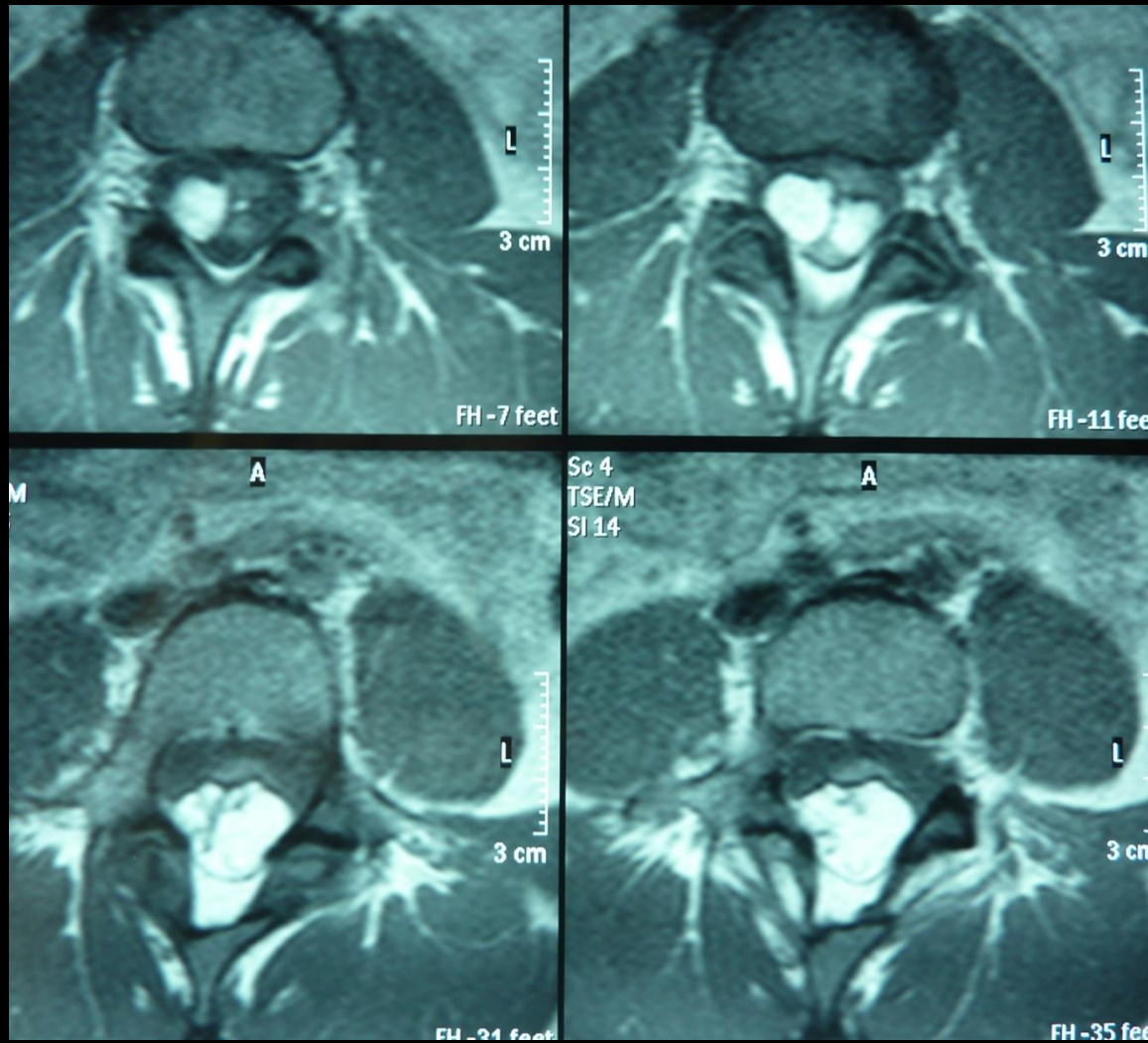
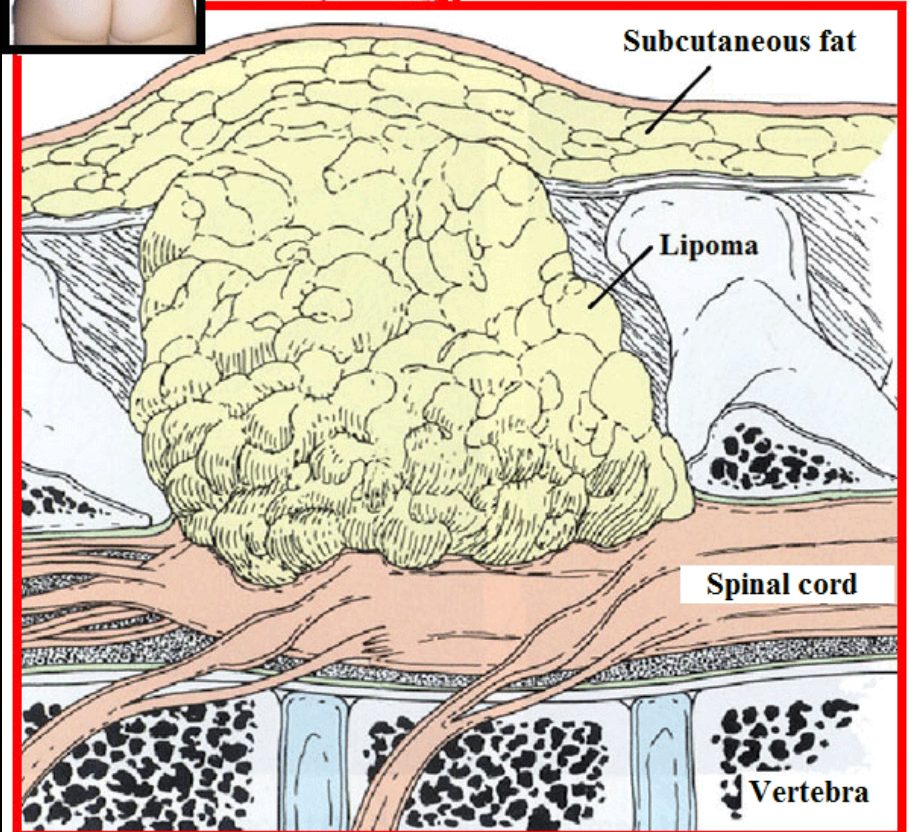
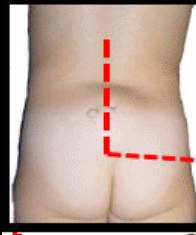
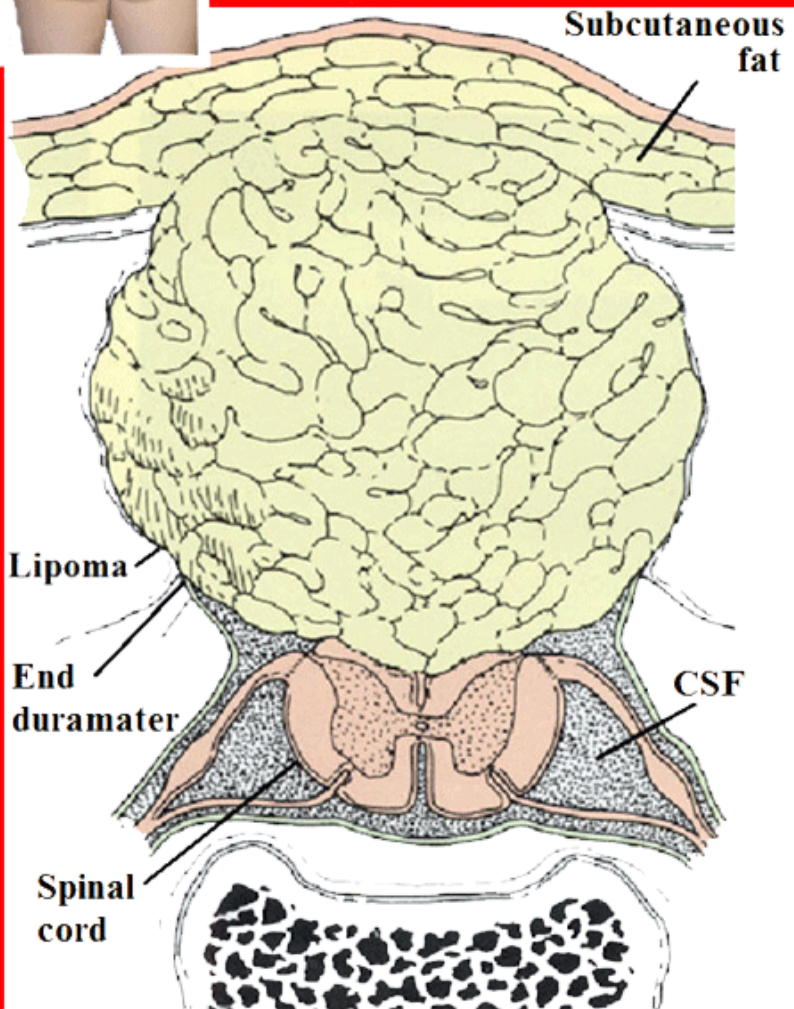
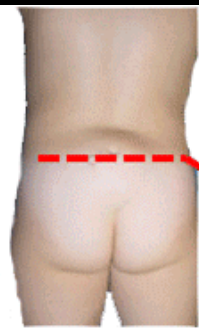
L
3 cm
FH -7 feet
L
3 cm
FH -11 fee
A
M
L
3 cm
Sc 4
TSE/M
SI 14
A
L
3 cm
FH -35 fee

Q.A., 25-year old female

- Urodynamics: mild detrusor hyperreflexia
- Hypoesthesia on the right foot
- Absent patellar reflex on the right

Lipoma del filum terminale

Lipoma del cono midollare

Lipoma del cono midollare

Subcutaneous fat
Lipoma
End duramater
CSF
Spinal cord

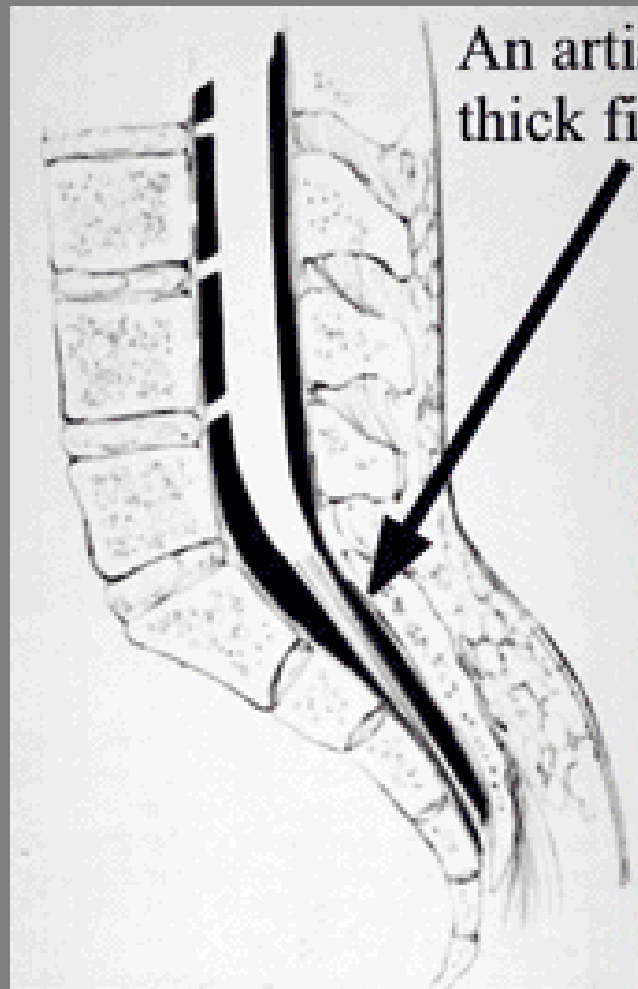
Subcutaneous fat
Lipoma
Spinal cord
Vertebra

Tethered cord syndrome and “normal level conus medullaris”

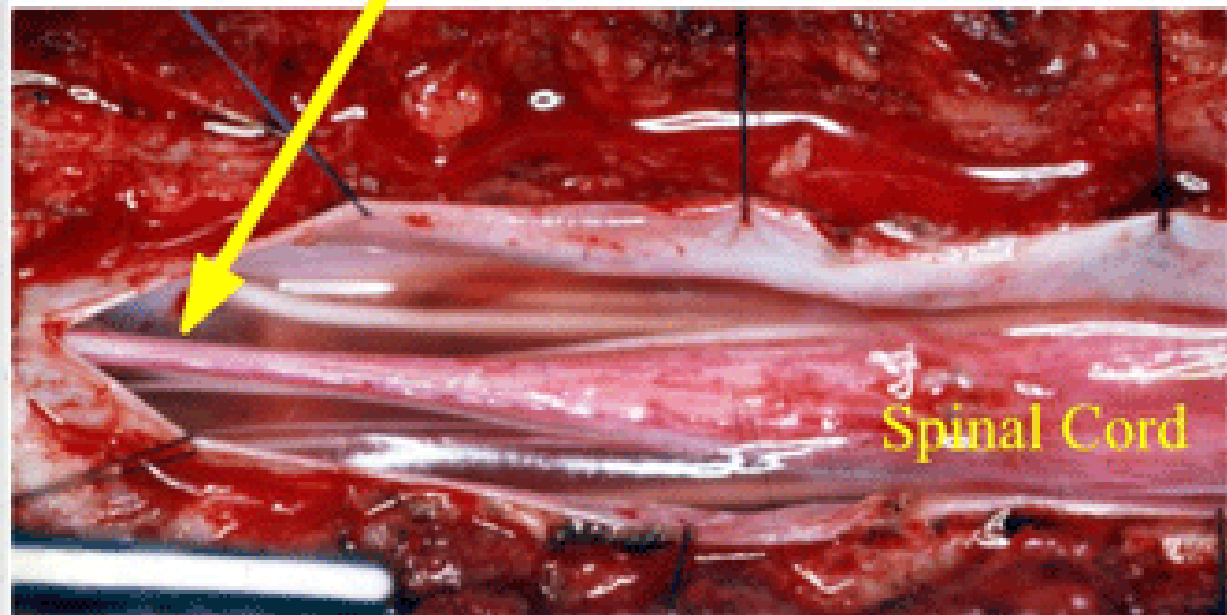
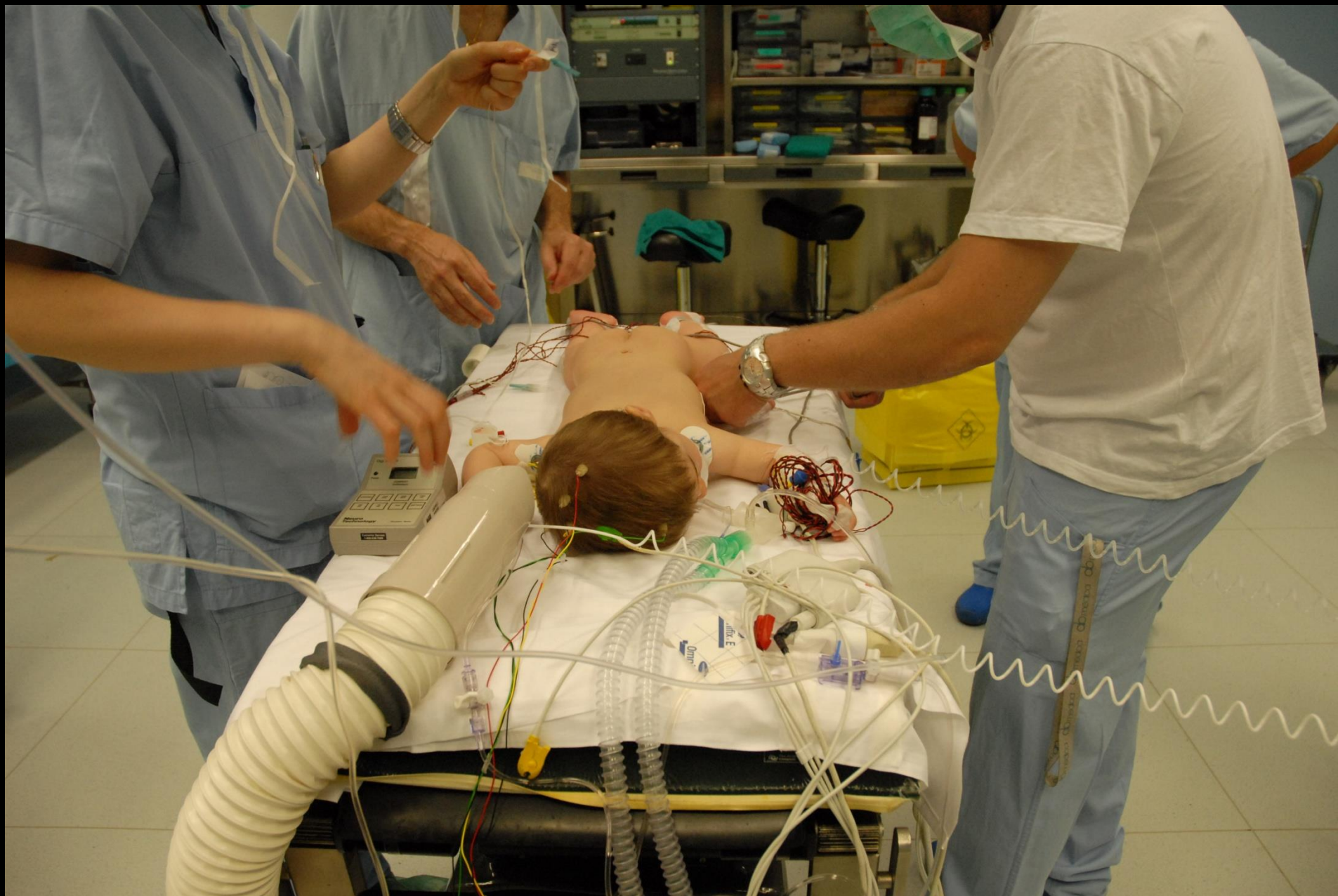
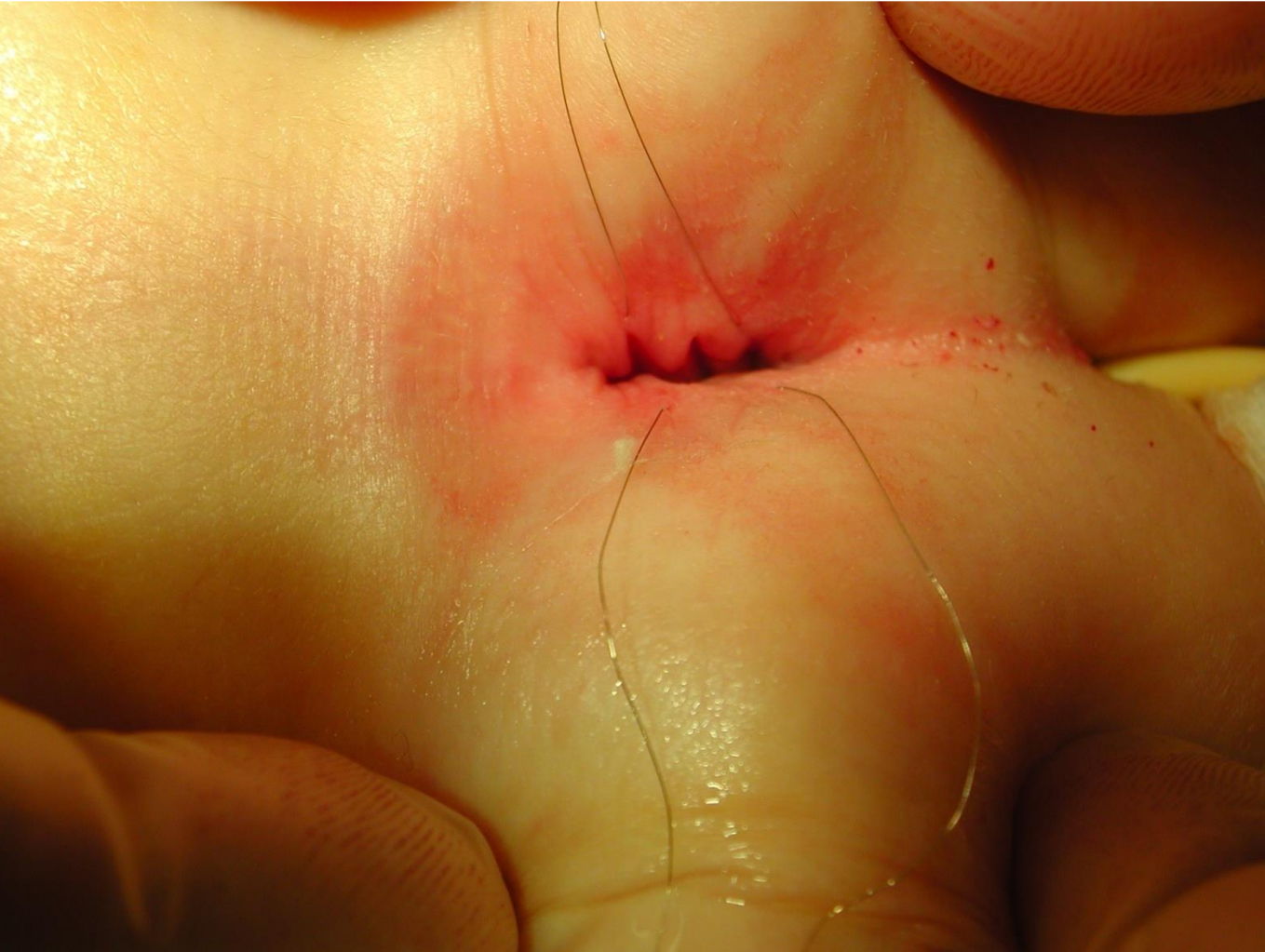
Conus medullaris

TCS and NLCM

- Hendrick et al. The tethered spinal cord. Clin Neurosurg 1983;30:457-463.
  - 12 out of 86 (14%)
- Raghavan et al. MR imaging in the tethered spinal cord syndrome. AJR 1989;152:843-852.
  - 4 out of 25 (16%)
- Warder et al. Tethered cord syndrome and the conus in a normal position. Neurosurgery 1994;34:597-600.
  - 13 out of 73 (18%)

Tethered cord

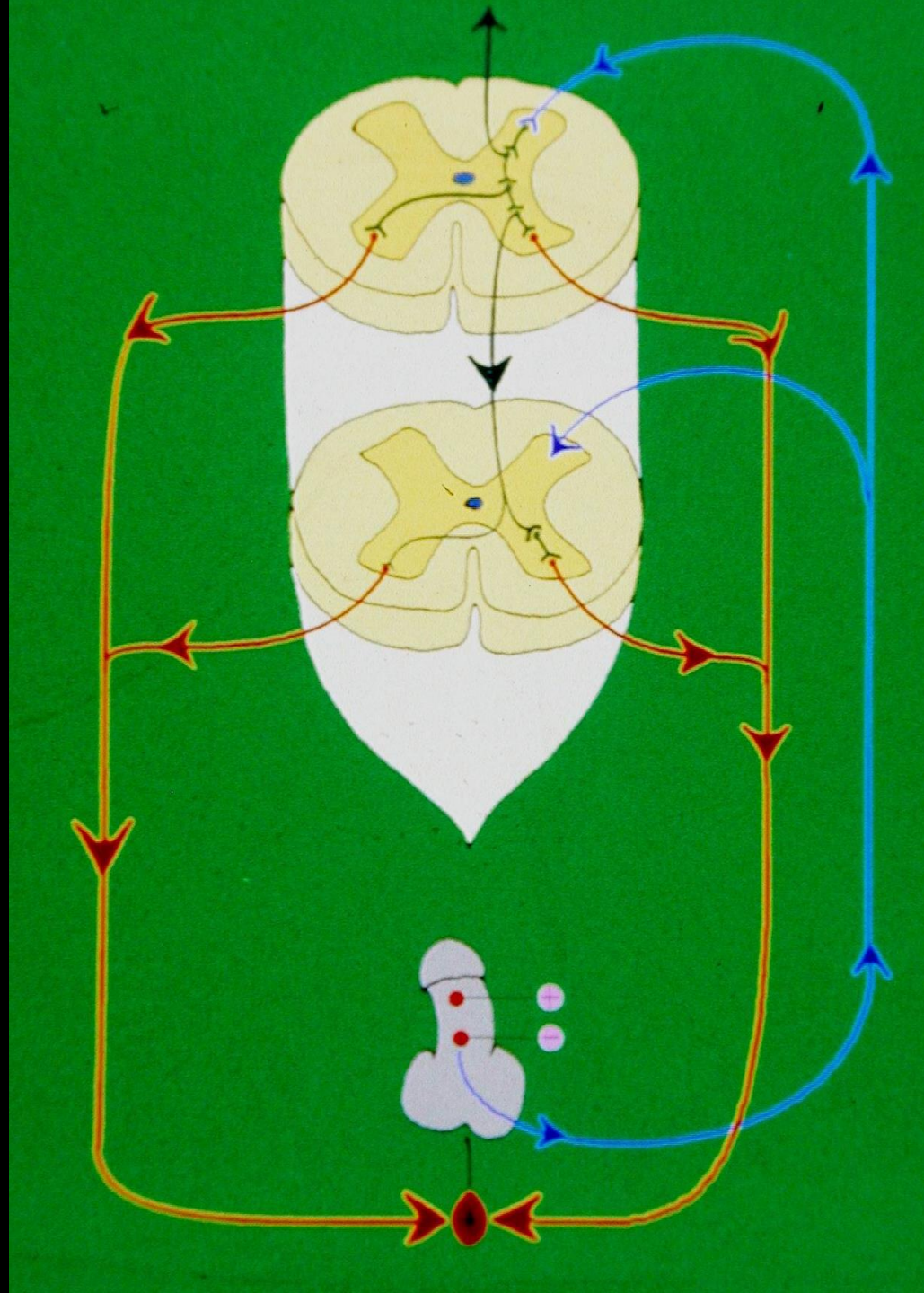
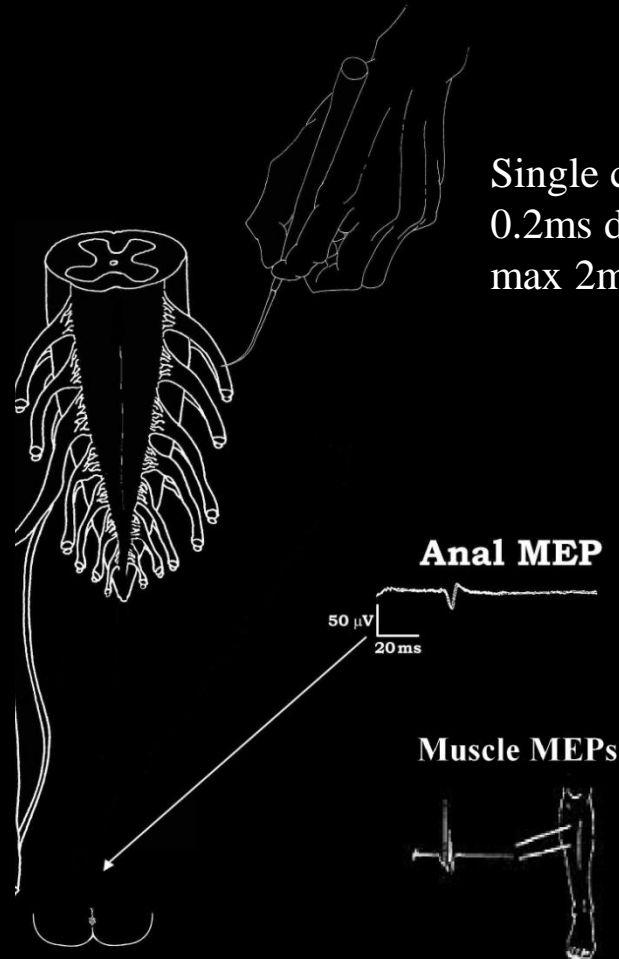
- Thick filum terminale
- Fatty filum terminale

An artist's drawing of a
thick filum terminale

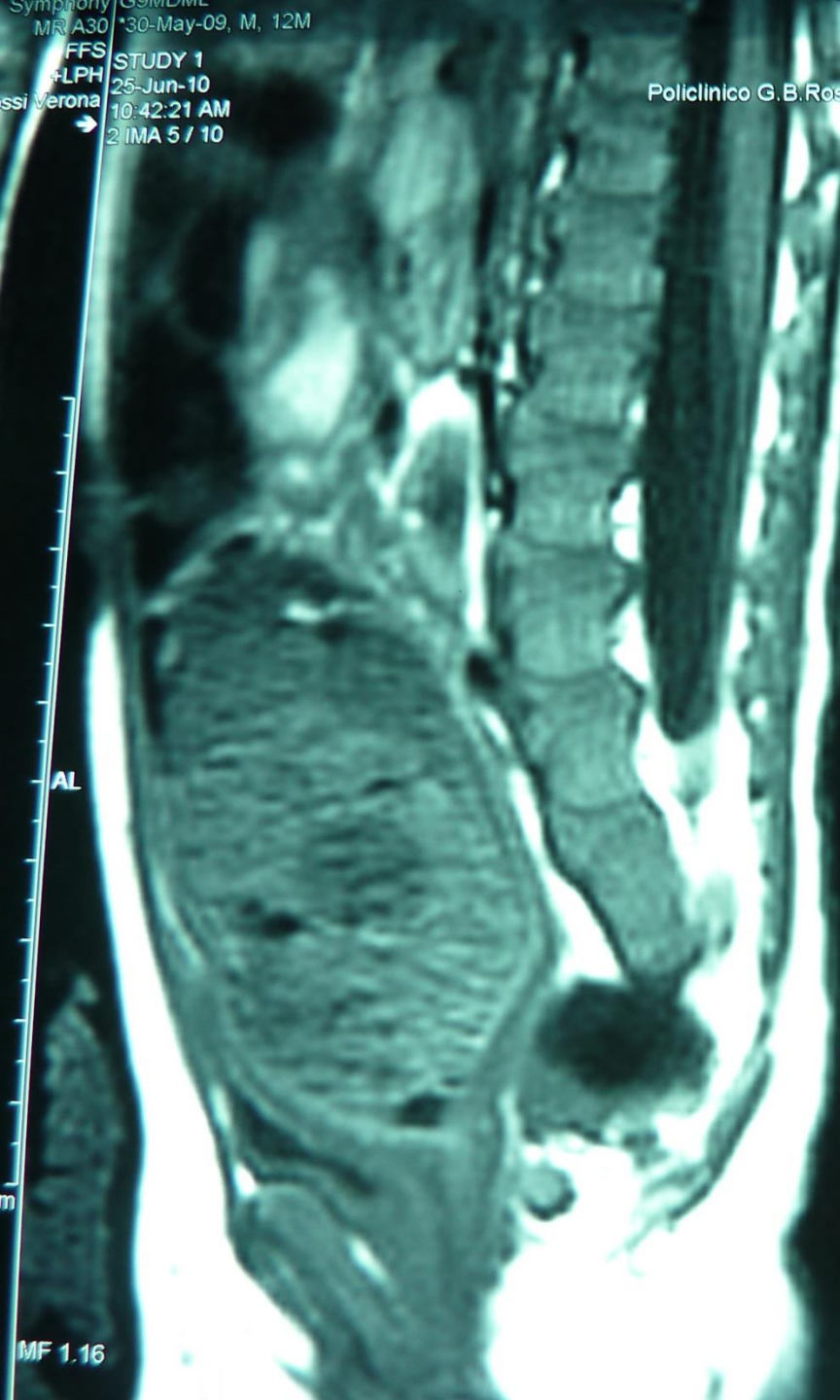
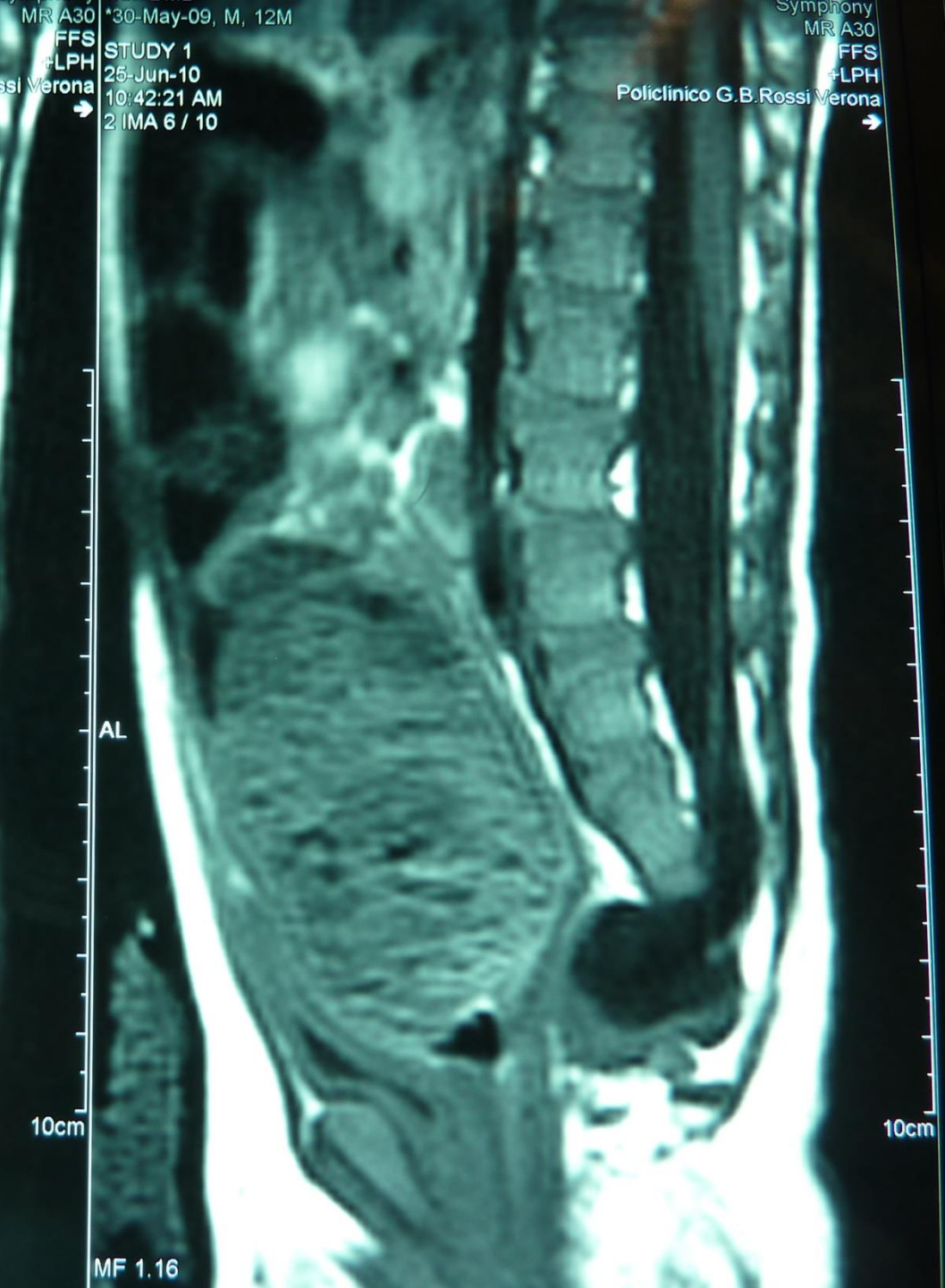
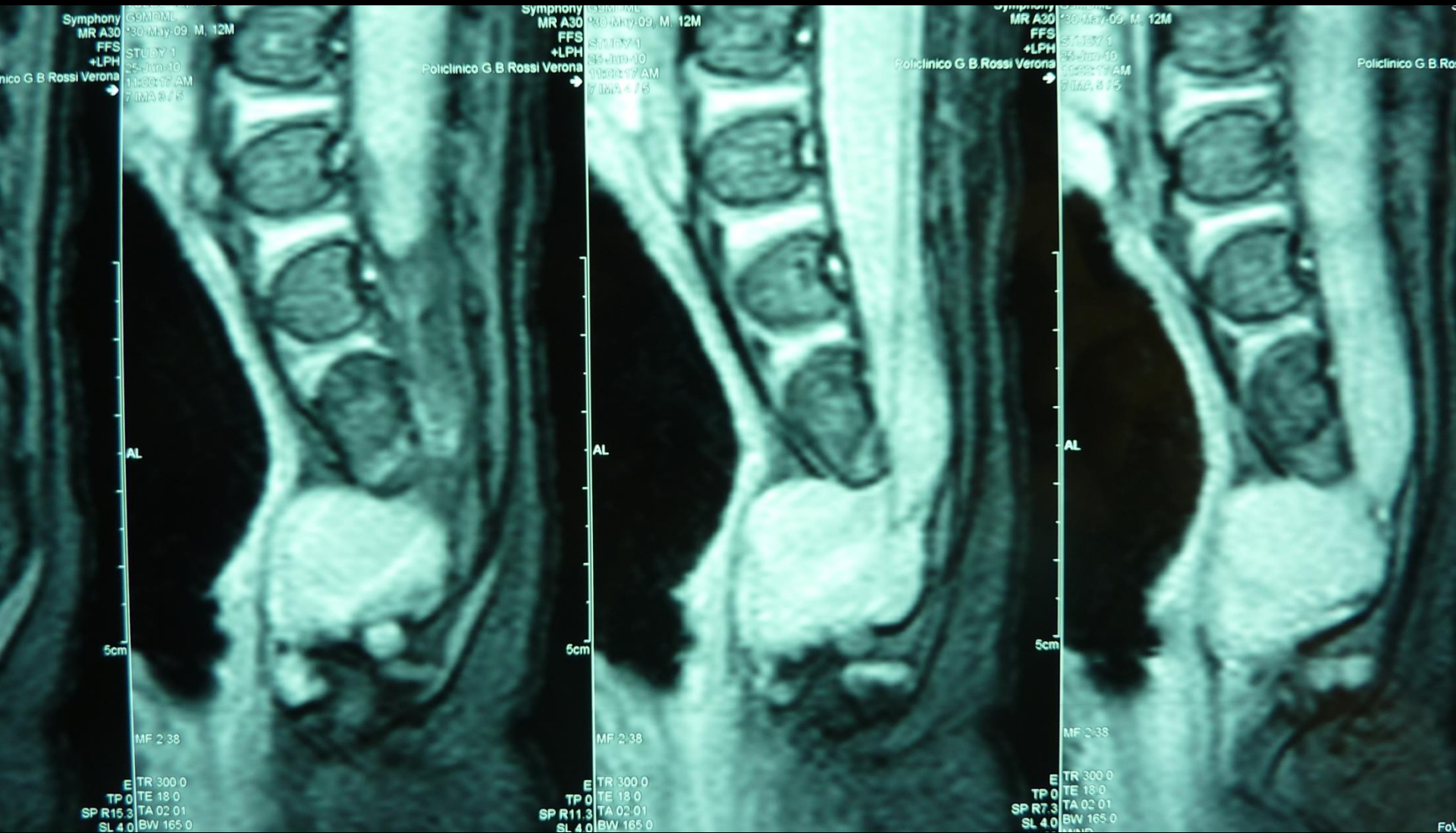
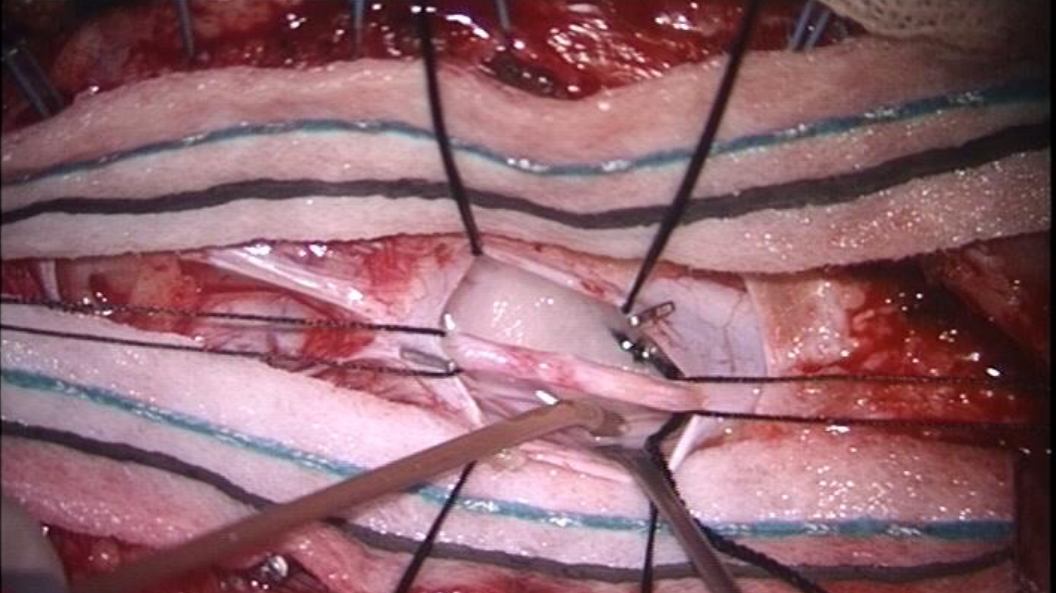
A surgical view of the thickened filum
Spinal Cord

The set-up

The bulbocavernosus reflex:

1) Oligosynaptic reflex

2) S2-S4 sacral segments

MOTOR MAPPING

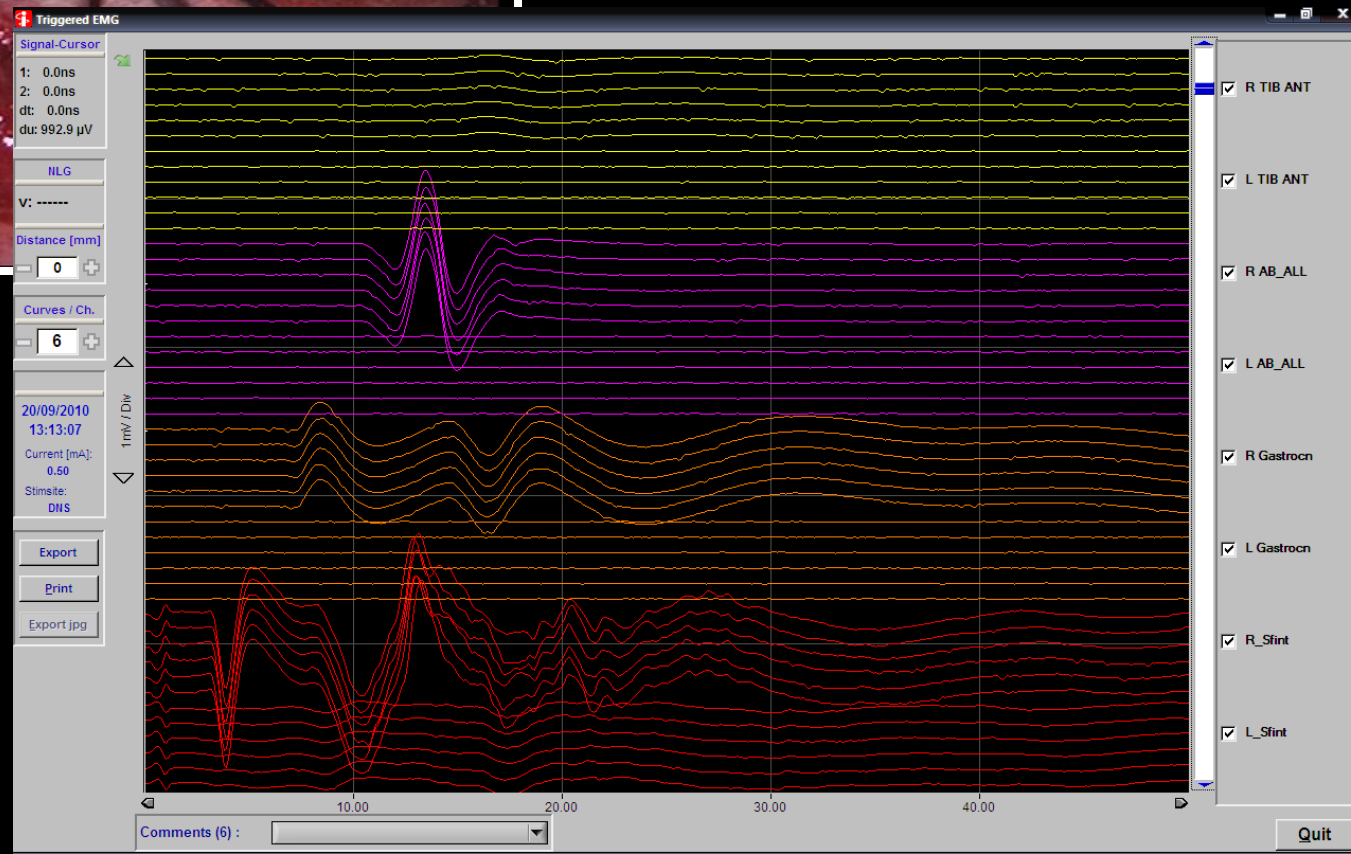
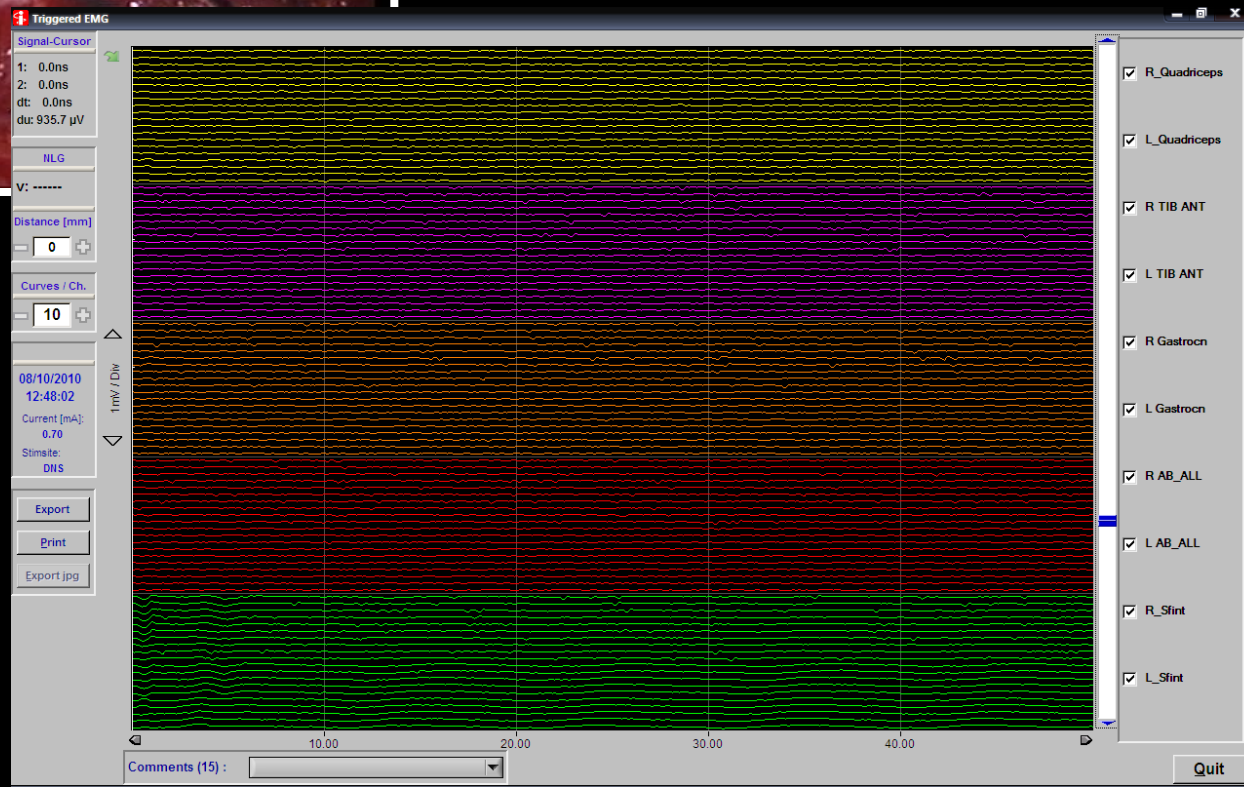
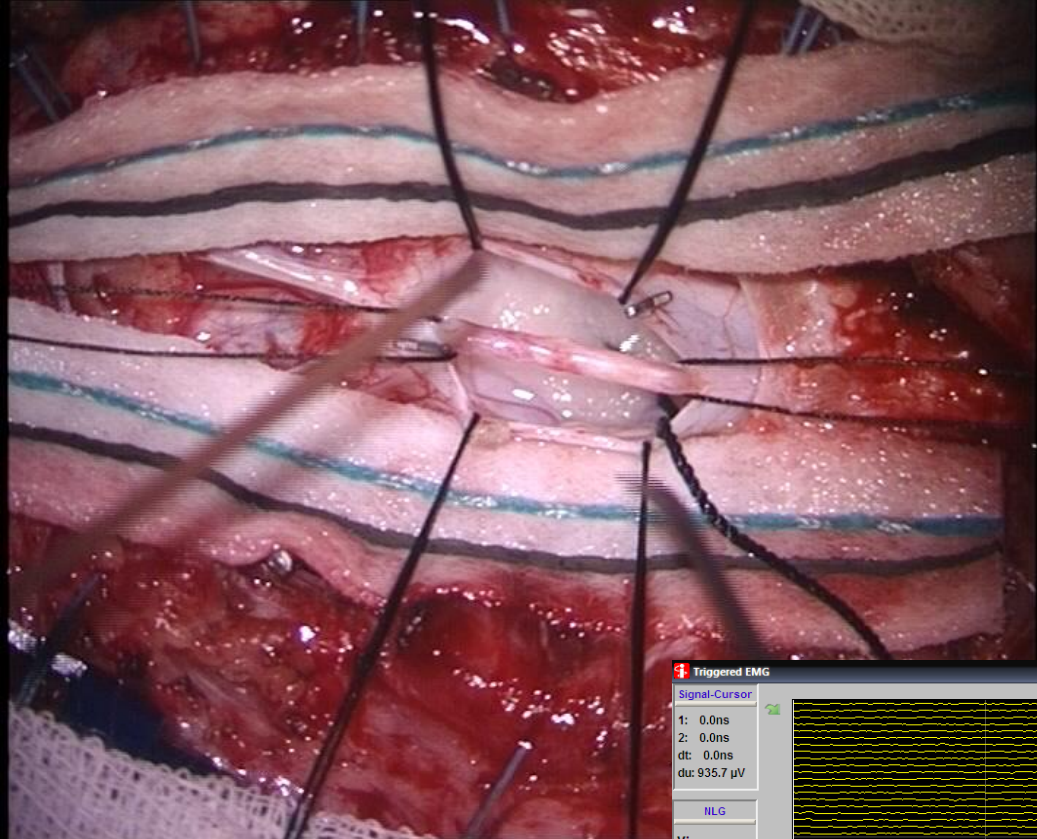
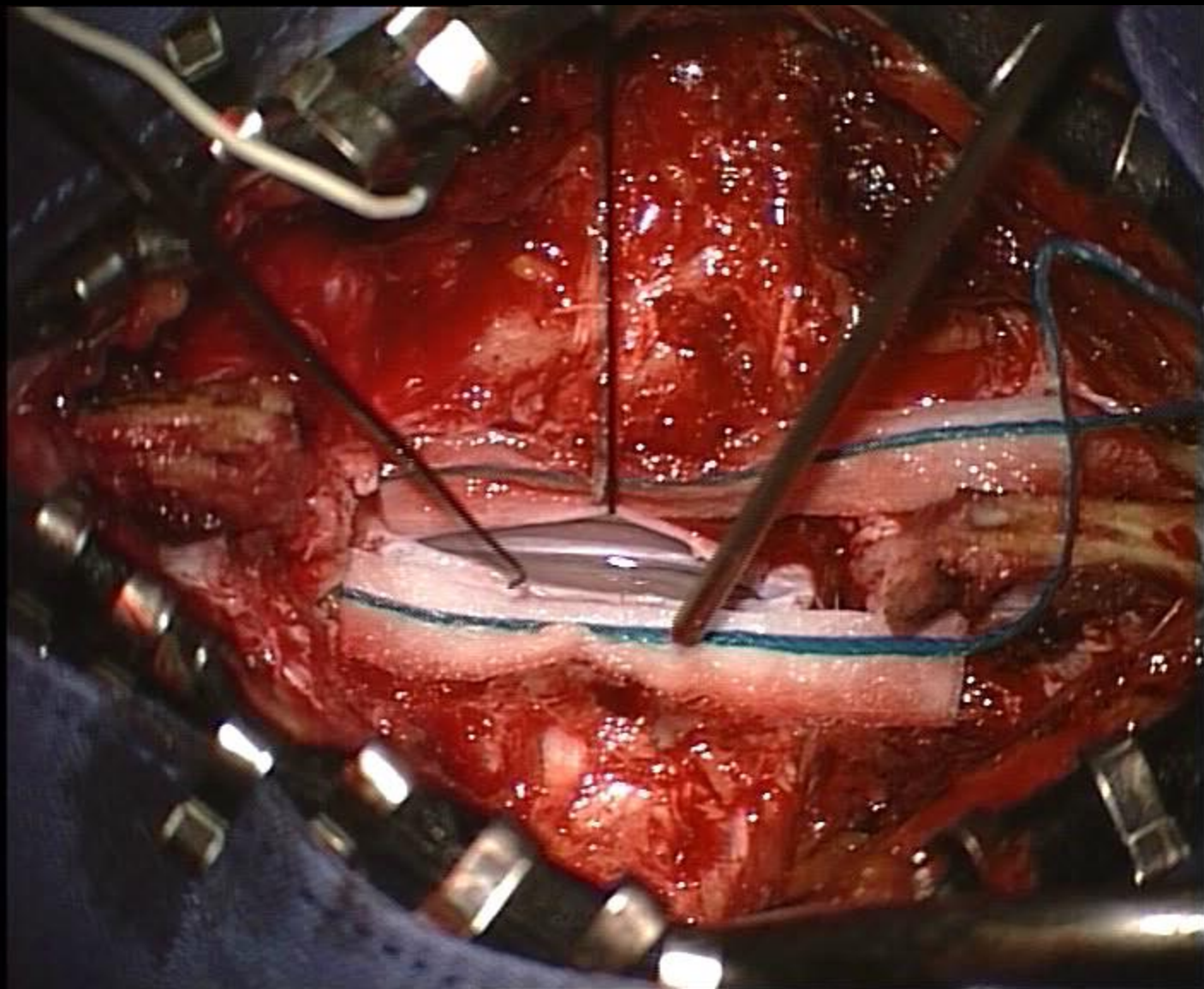
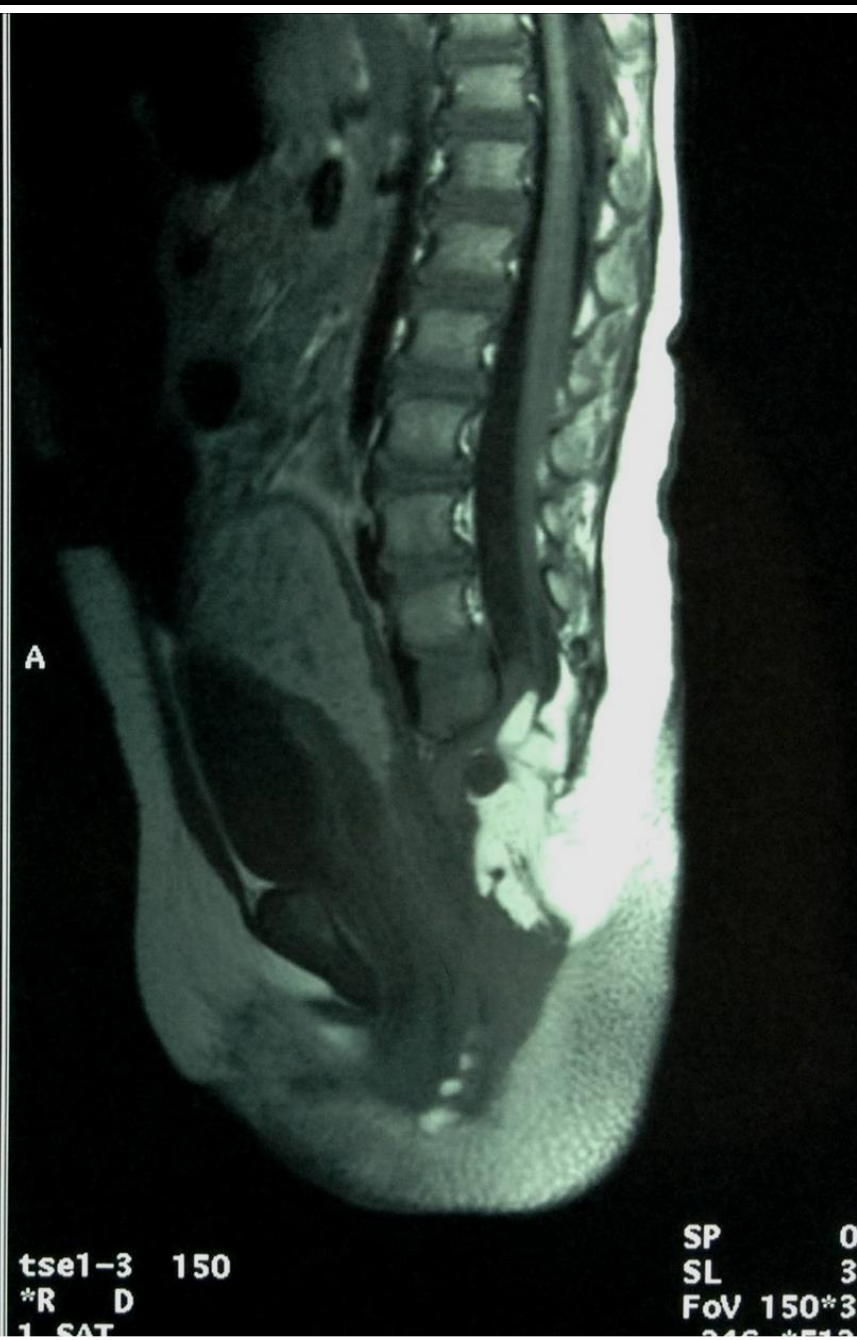
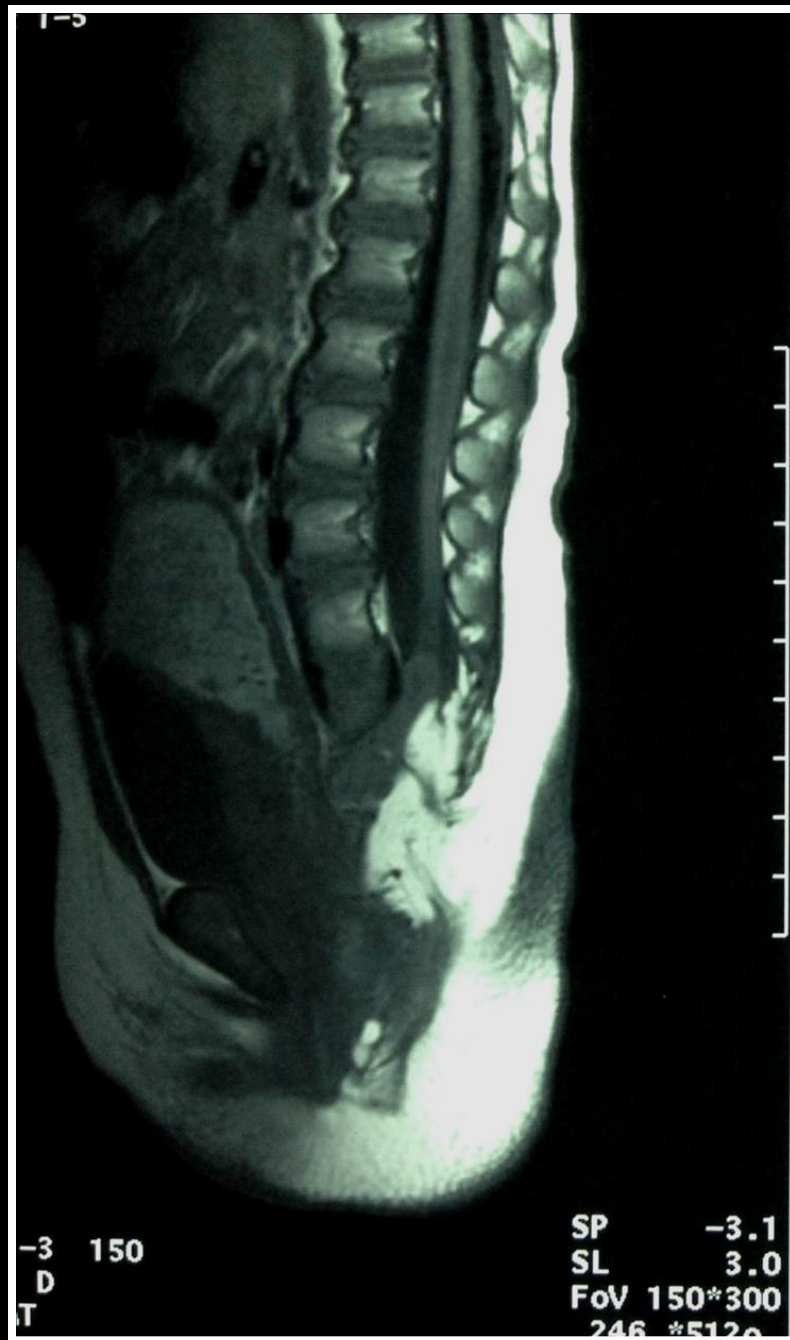
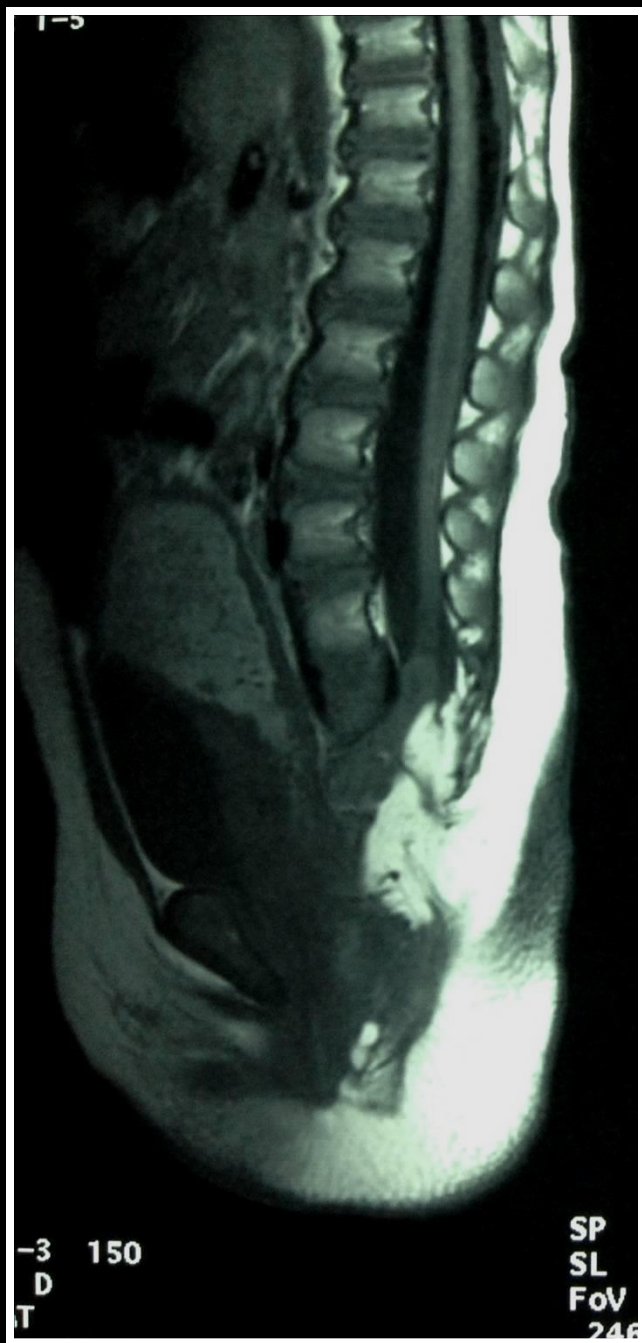
Single cathodal stimulation:
0.2ms duration
max 2mA intensity

Motor mapping for lumbosacral roots

	L4	L5	S1	S2	S3	S4
Quadriceps (L2-L4)						
Tibialis anterior MEPs						
Gluteus Maximus						
Abductor hallucis brevis MEPs Gastrocnemius						
Anal sphincter MEPs (efferent BCR)						

Anterior meningocele with tethered cord

Intensity 0.5 mA, right side m
Triggered EMG
Signal-Cursor
1: 0.0ns
2: 0.0ns
dt: 0.0ns
du: 992.9 µV
NLG
V: ------
Distance [mm]
0
Curves / Ch.
6
20/09/2010
13:13:07
Current [mA]:
0.50
Stimsite:
DNS
Export
Print
Export jpg
1mV / Div
R TIB ANT
L TIB ANT
R AB_ALL
L AB_ALL
R Gastrocn
L Gastrocn
R_Sfint
L_Sfint
10.00
20.00
30.00
40.00
Comments (6) :
Quit

-3 150
D
SP -3.1
SL 3.0
FoV 150*300
A
tse1-3 150
*R D
SP 0
SL 3

-3
150
D
SP
SL
FoV

A
Pang 2011

B
Pang 2011

Retained Medullary Cord in Humans: Late Arrest of Secondary Neurulation

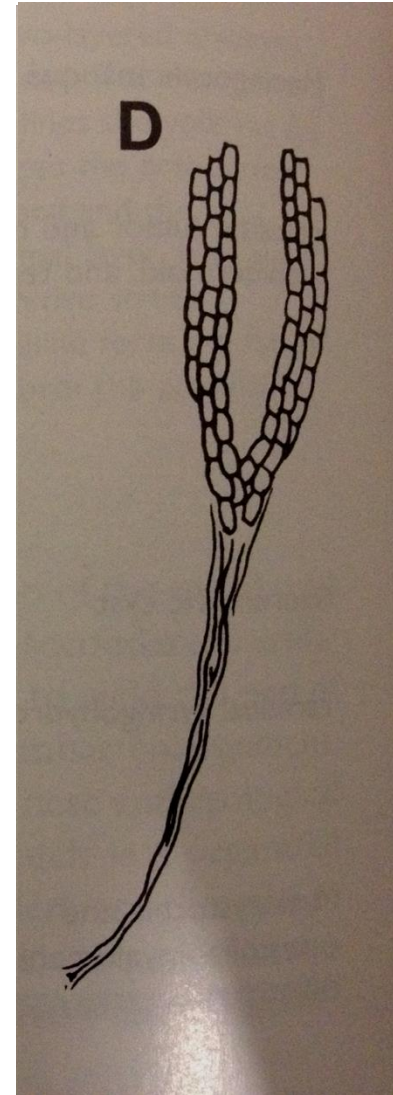
Dachling Pang, MD, FRCS(C),FACS*‡
John Zovickian, MD, FACS‡
Greg S. Moes, MD‡§¶

BACKGROUND: Formation of the caudal spinal cord in vertebrates is by secondary neurulation, which begins with mesenchyme-epithelium transformation within a pluripotential blastema called the tail bud or caudal cell mass, from thence initiating an event sequence proceeding from the condensation of mesenchyme into a solid medullary cord, intrachordal lumen formation, to eventual partial degeneration of the cavitatory

We encountered 7 patients who possessed an elongated central neural structure caudal to and in seamless continuation with the functional conus, complete with nerve roots distal to the last functional anal sphincter (S_4) roots.

Intraoperative motor mapping and histopathological findings suggest this aberrant neural structure may be a cavitary medullary cord (secondary neural tube) arrested in development just before its programmed destruction. This entity is of clinical interest because 5 of the 7 medullary cords acted as a tethering lesion.

Secondary Neurulation

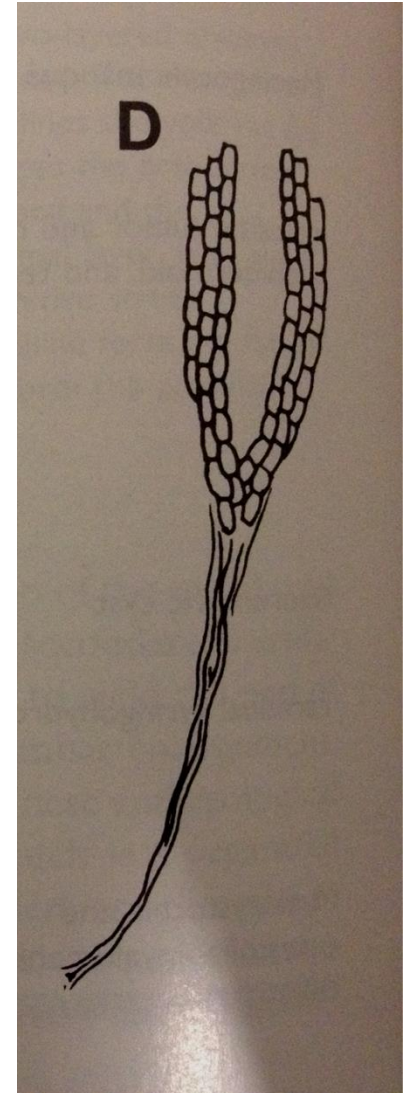
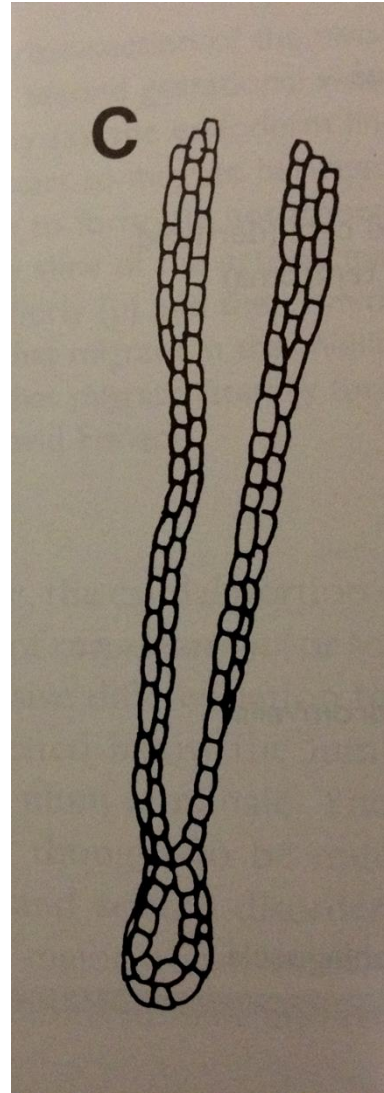
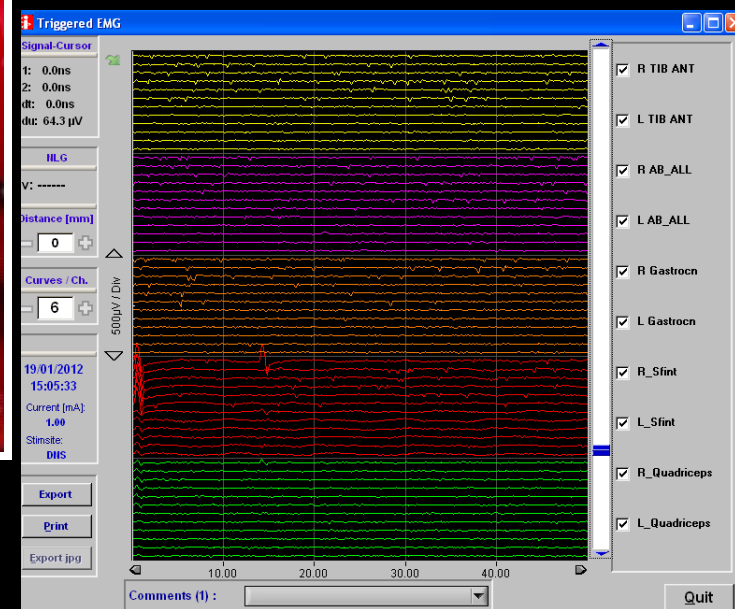
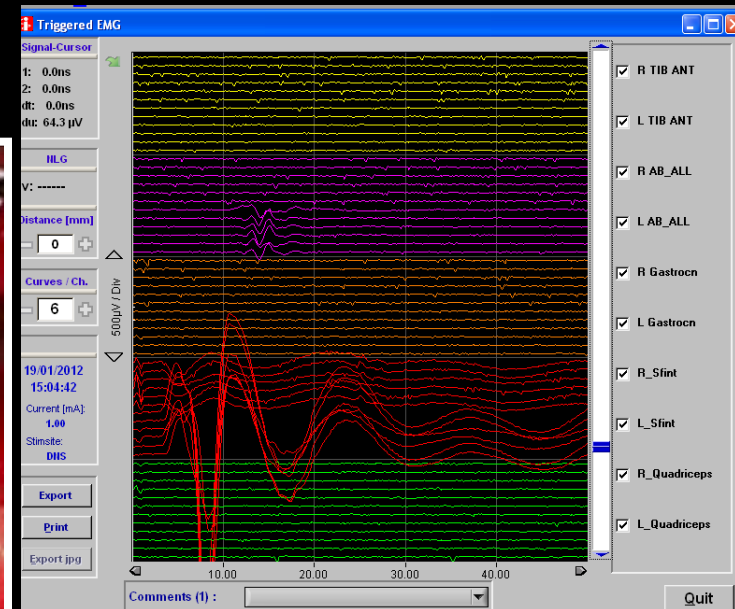
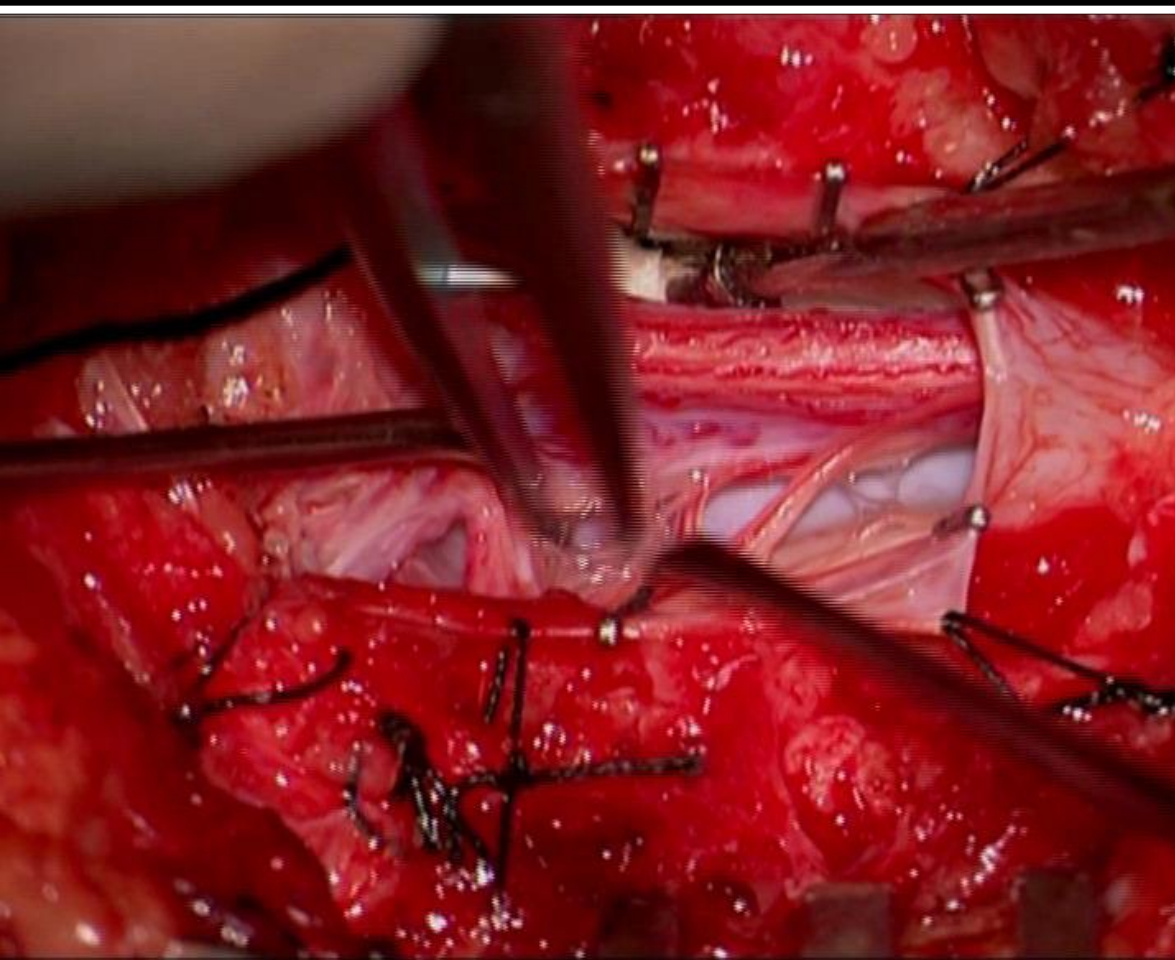
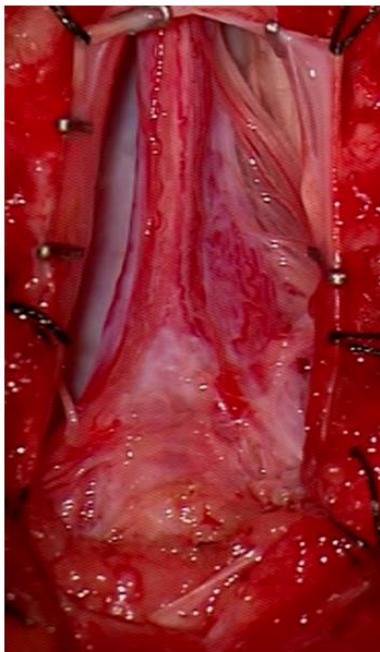
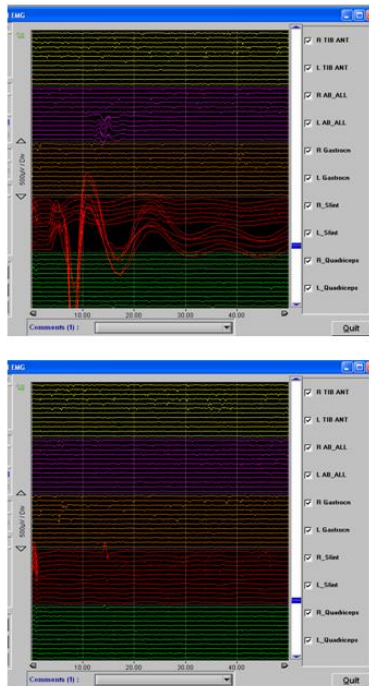
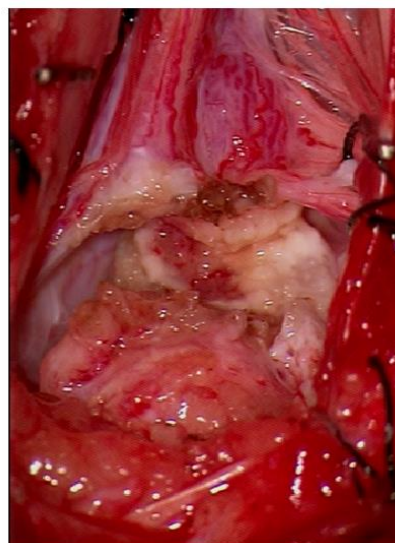
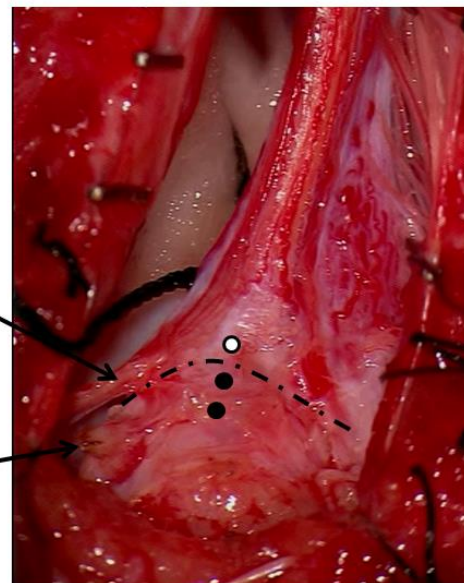
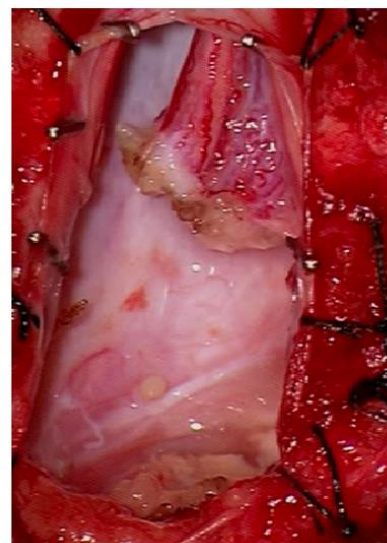
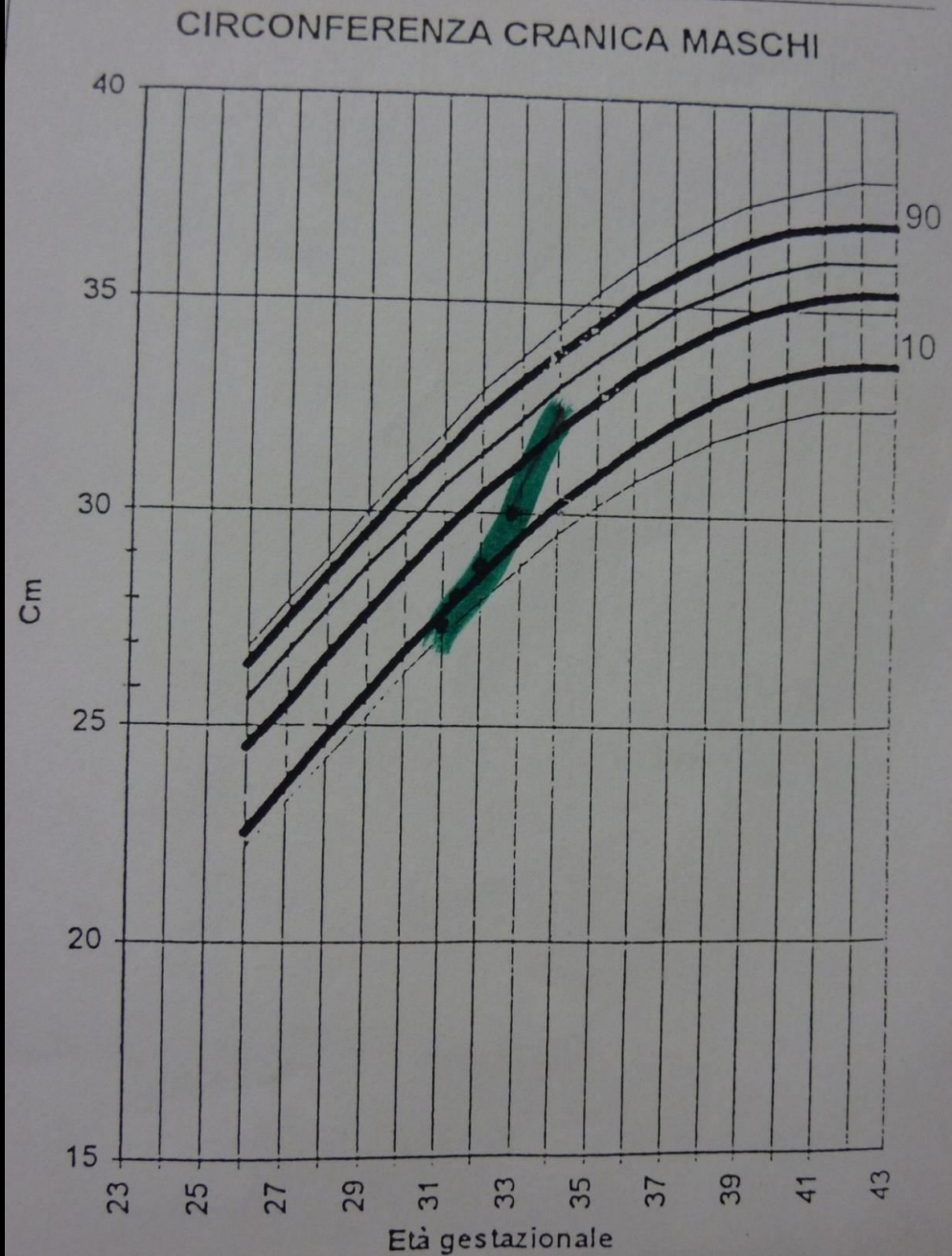
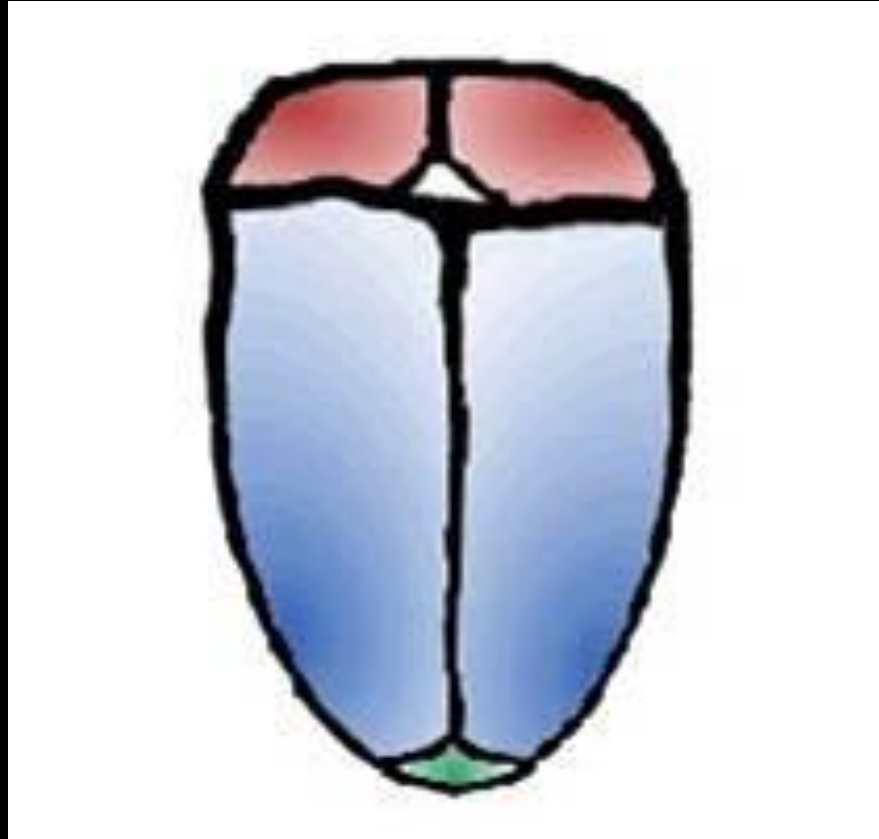
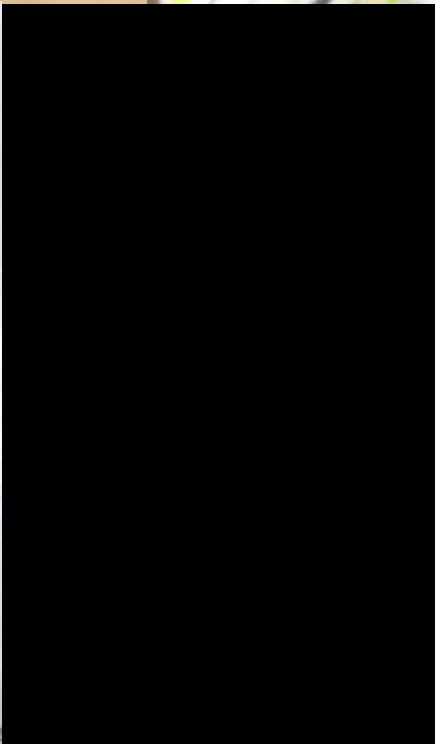
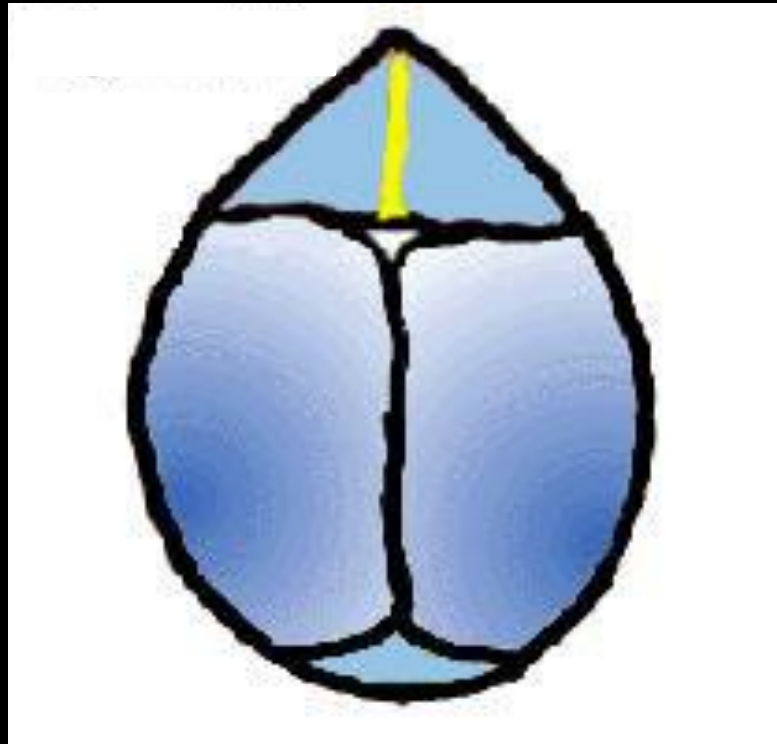
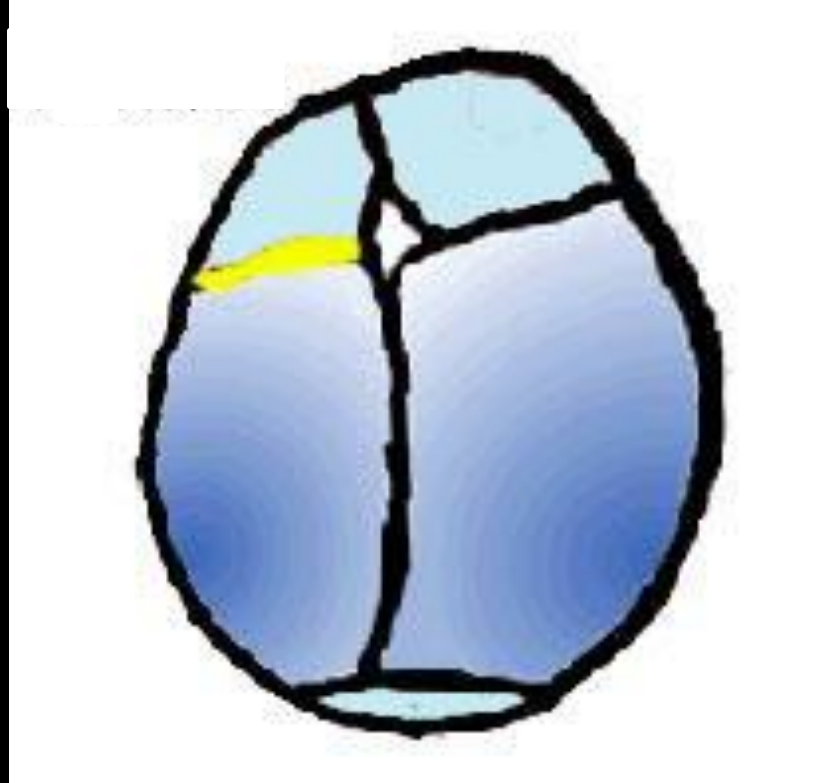
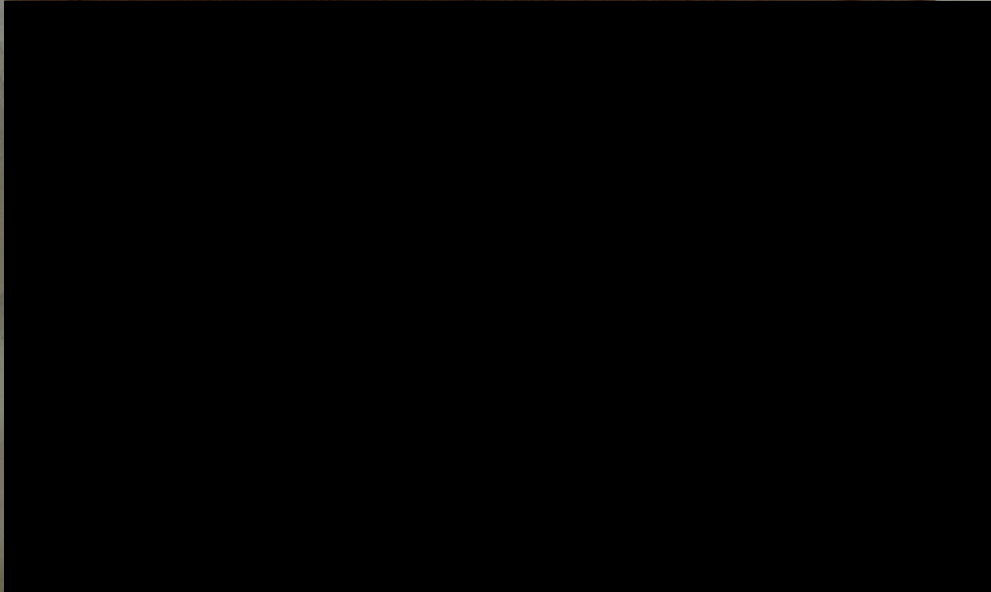
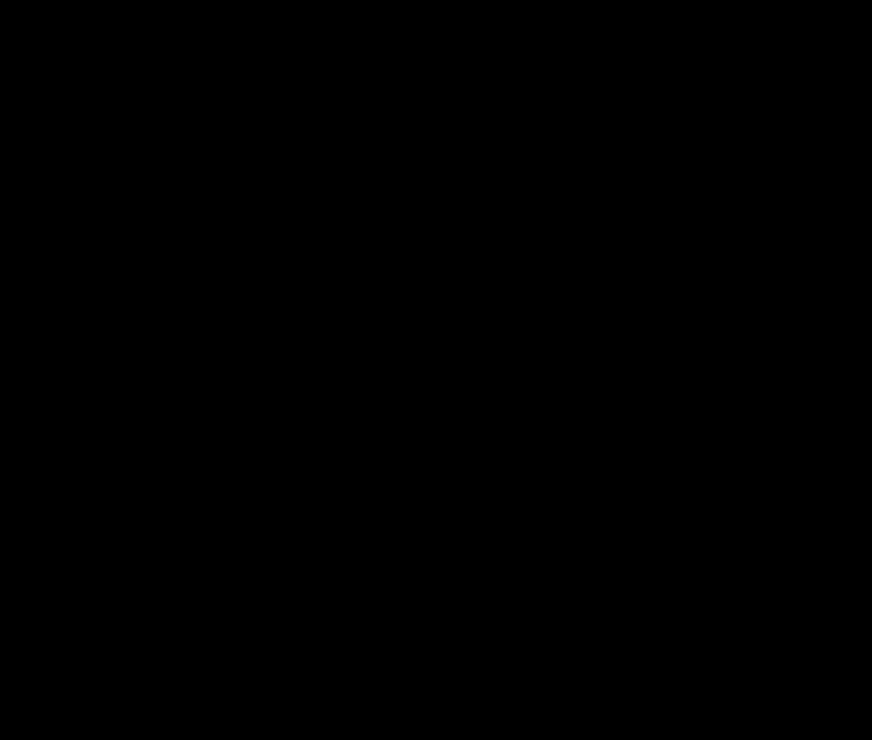
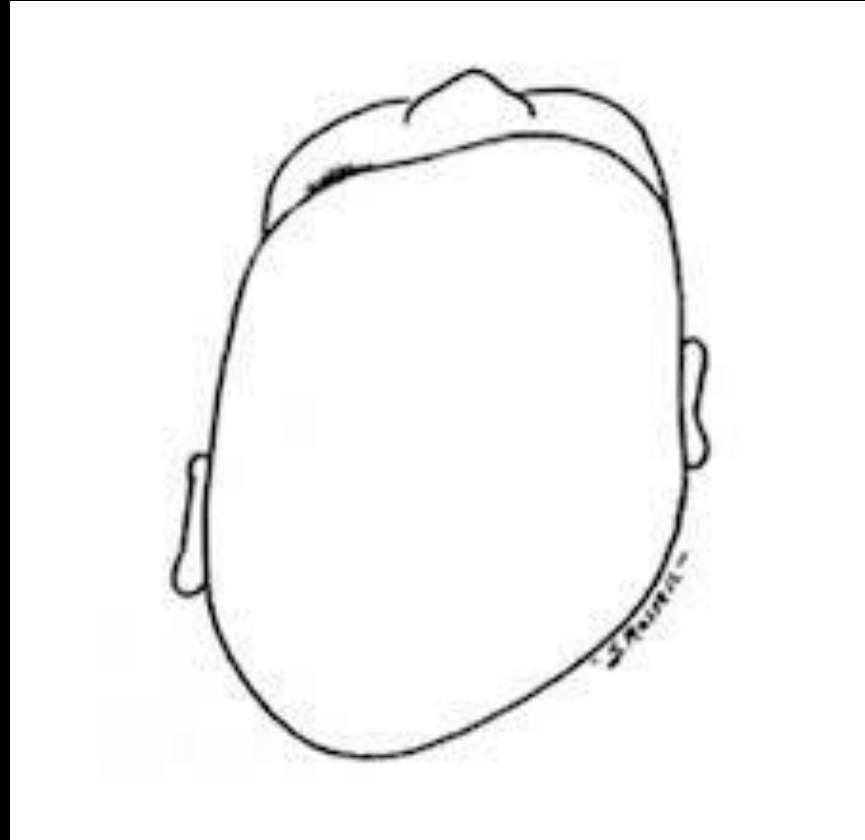
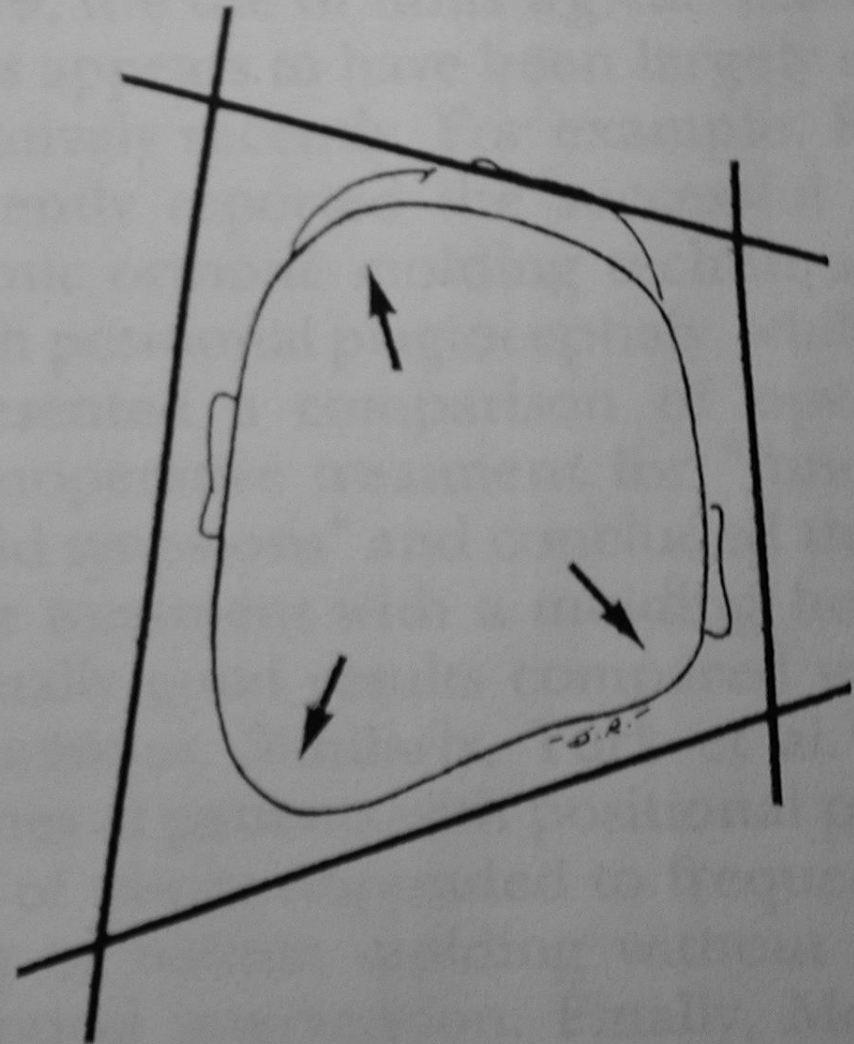
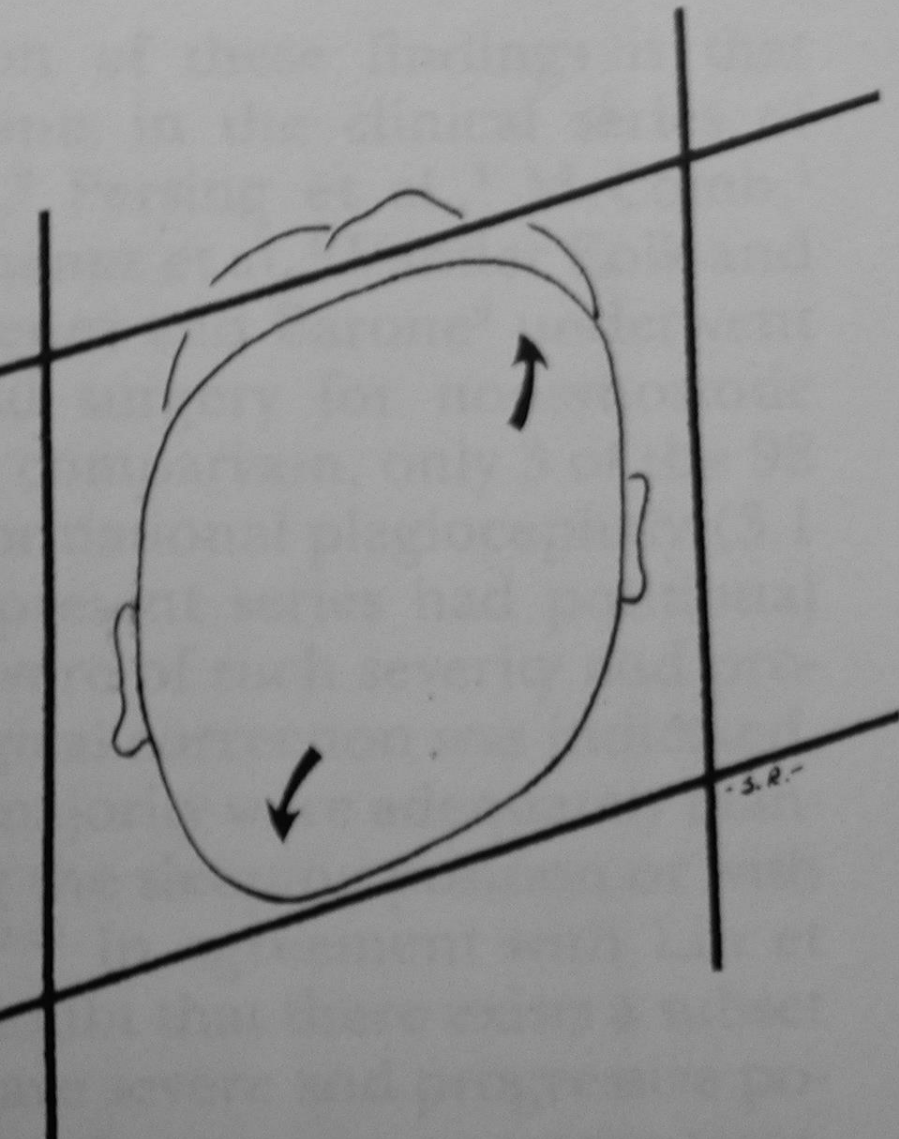
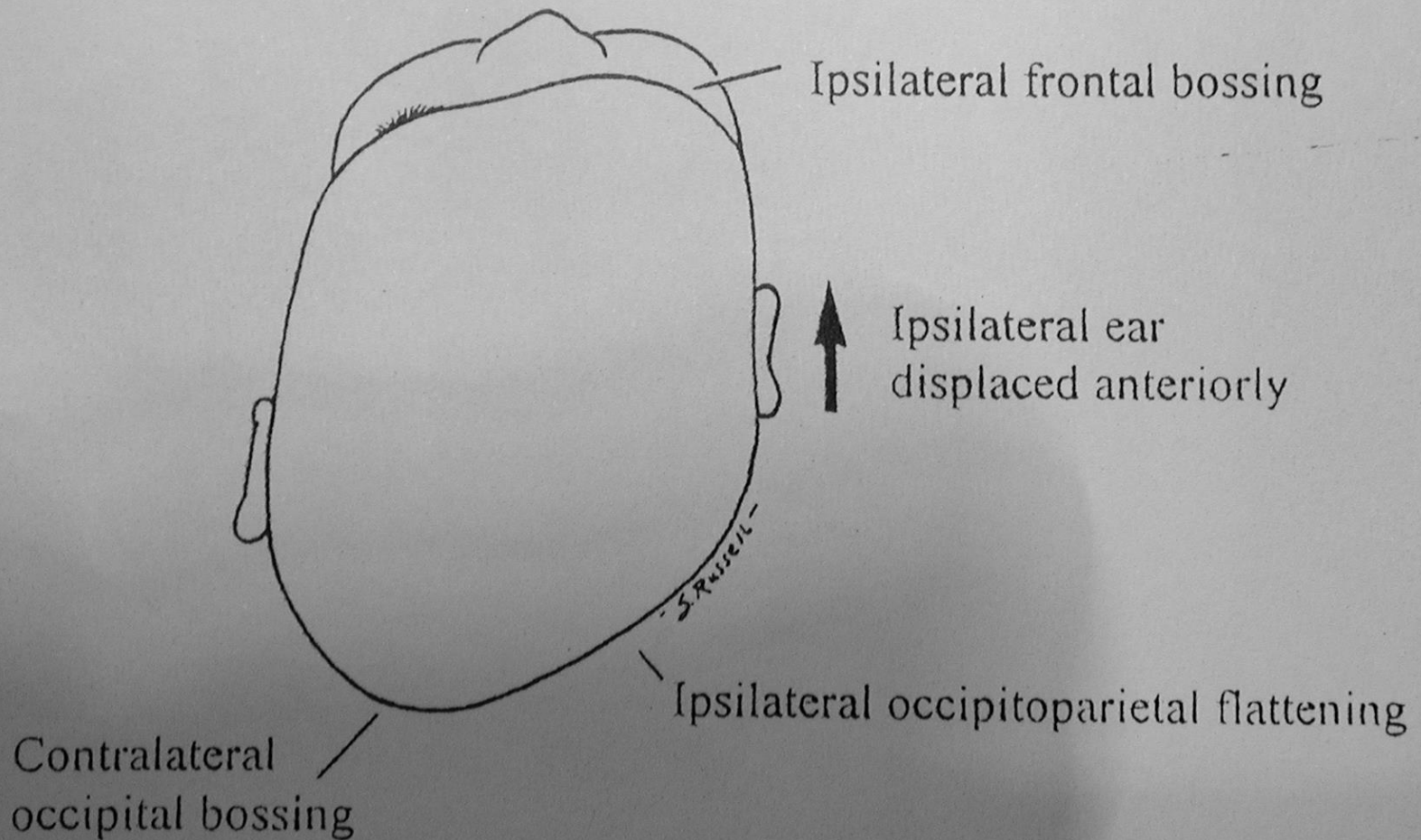
- Cellular aggregation and condensation to form the medullary cord
- Cavitation of the medullary cord
- Partial regression and degeneration of the cavitary medullary cord

Secondary Neurulation

- Cavitation of the medullary cord
- Partial regression and degeneration of the cavitary medullary cord

?

CAROSELLO

Appello…

CIRCONFERENZA CRANICA MASCHI
40
35
30
25
20
15
Cm
90
10
23
25
27
29
31
33
35
37
39
41
43
Età gestazionale

Scafocefalia

Trigonocefalia

Plagiocefalia anteriore

Plagiocefalia posteriore

Plagiocefalia posteriore

Posizionale

Sinostosi lambdoidea

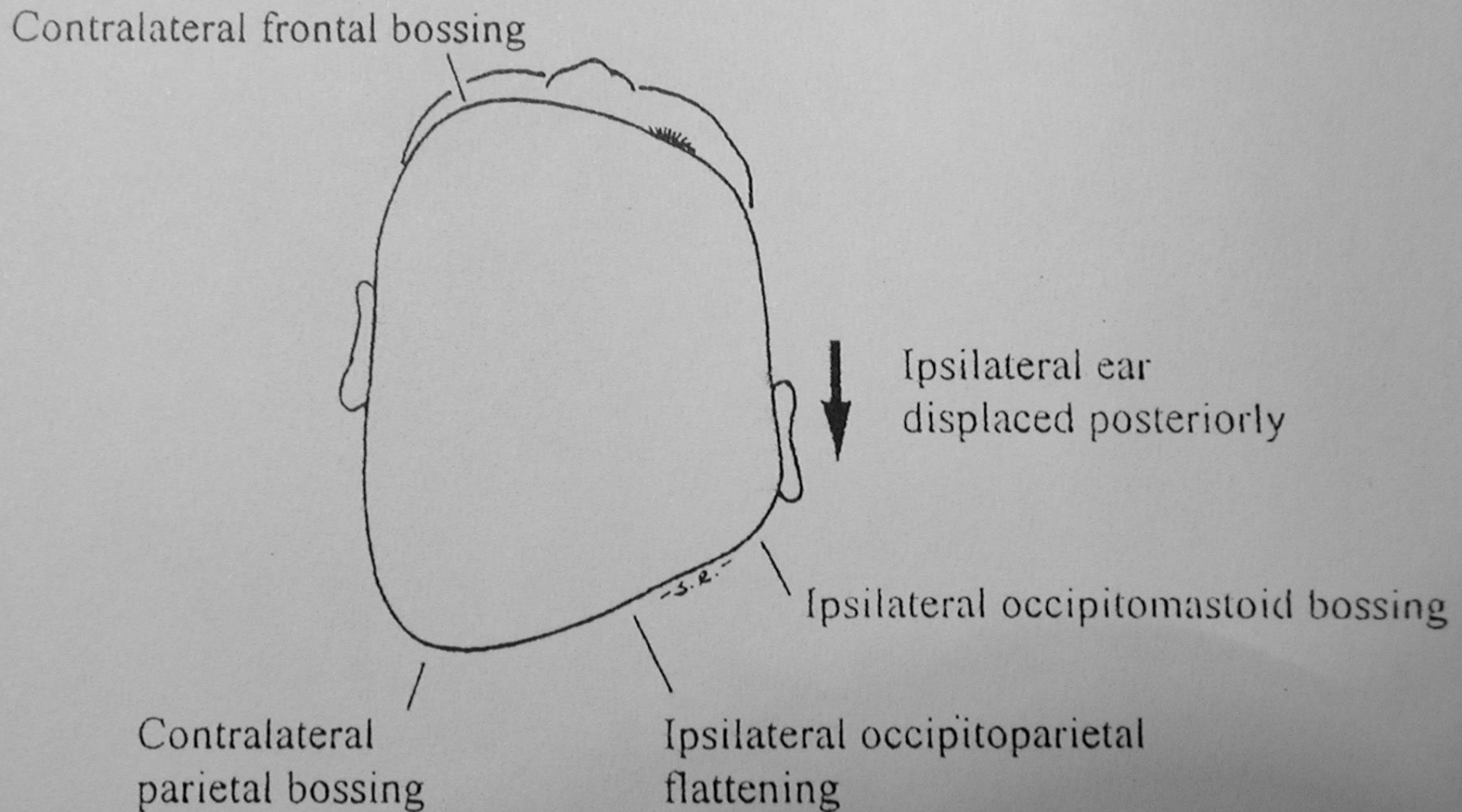
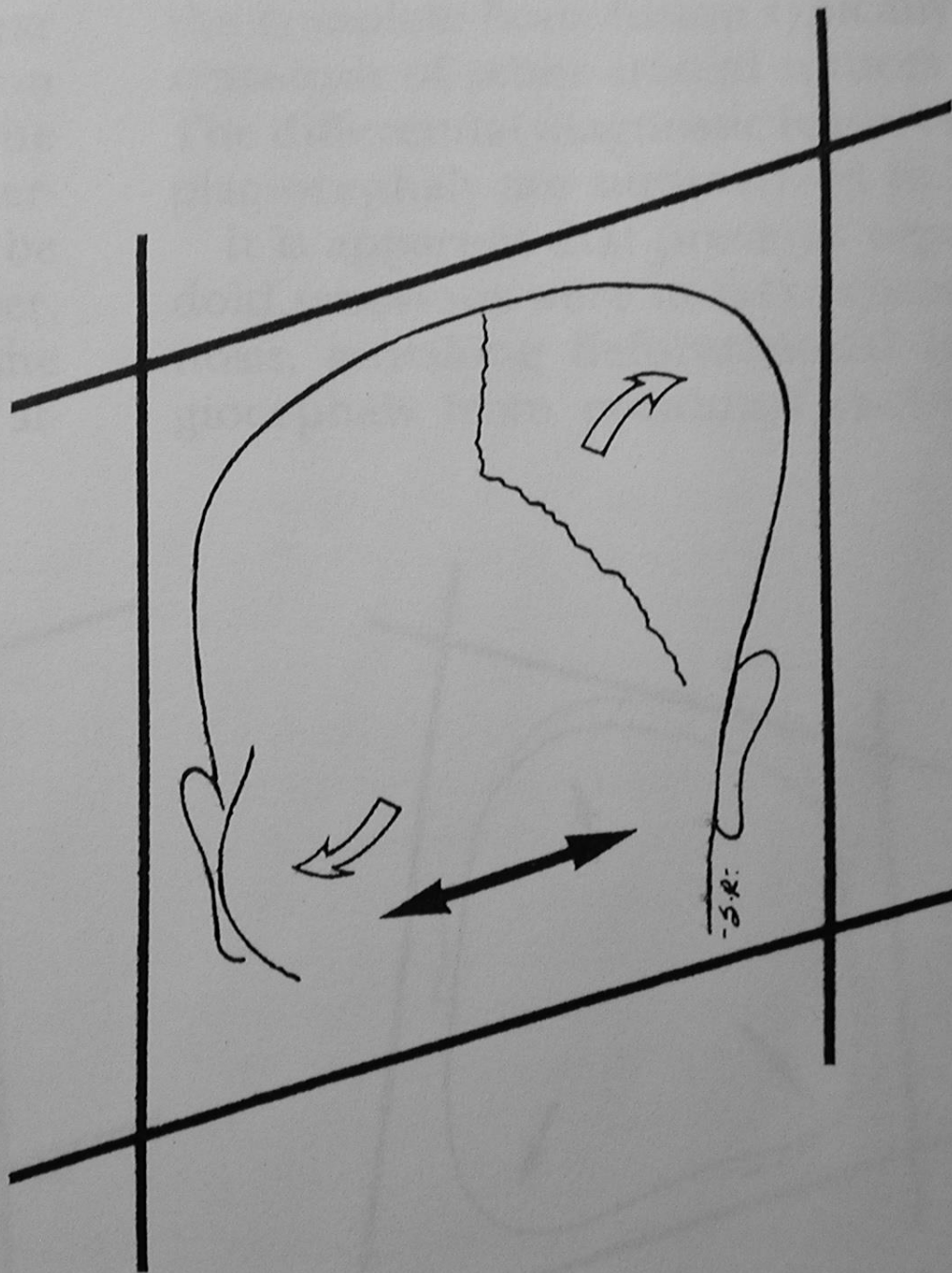
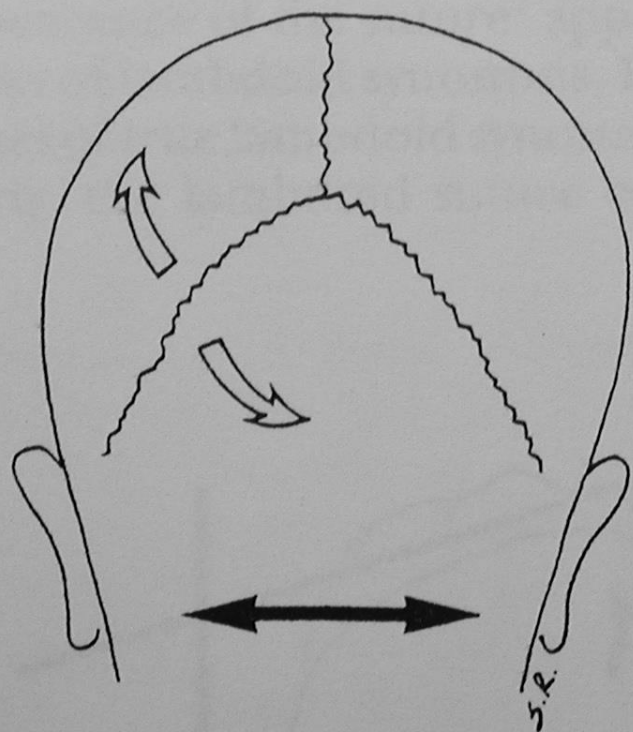



FIG. 10. Differences between deformational and synostotic posterior plagiocephaly from

Plagiocefalia posizionale

- E’ dovuta ad una preferenza posturale del bambino
- Può essere associata a torcicollo
- Più frequente in infanti con scarsa attività motoria, maschi, primogeniti
- In aumento dopo il consiglio al decubito supino per prevenzione della morte in culla
- Incidenza fino a 1/68

Plagiocefalia posizionale

- Entro i 6 mesi, solo consigli sulla postura
- Dopo i 6 mesi alcuni consigliano il caschetto (?)
- Chirurgia anedottica
- Fino ai 12 mesi ci si può attendere un beneficio dalla postura

francesco.sala@univr.it